ABSTRACT

Objective: Studies and professional organizations have promoted genetic counseling for individuals with sickle cell disease (SCD) and SCD carrier status. Because genetic testing within the sickle cell context has historically been tensioned by racial discrimination and stigma in the U.S., this study explored current opinions and experiences from stakeholders to inform ways to enhance the benefits and mitigate perceived harms of genetic counseling in the sickle cell context. We also queried perspectives on racialized versus ancestral views of SCD in the medical and broader communities.

Methods: Five focus group discussions were conducted with 21 adults who have SCD. Open-ended questions and probes covered topics including experiences living with SCD, experiences with and perceptions of genetic counseling, perspectives on racialized views and ancestral framing of SCD. Transcribed discussions were coded and interpreted through inductive thematic analysis within a critical realism methodological paradigm.

Results: Fifteen themes from the data were organized under four sections:

Section (A) “Experiences in the Personal, Social, and Healthcare Contexts' highlights common experiences of individuals with SCD. In the healthcare context, participants expressed seeking partnership with healthcare professionals, stressing that effective communication as a key factor to collaboration. Barriers were explained to interfere with interactions and partnership goals. Individuals also described a low awareness of SCD in both the medical and greater communities, which can be consequential for those who live with the disorder.
Section (B) “Dimensions of Decision Making” illustrates various psychological and social challenges men and women with sickle cell experience when making reproductive decisions.

Section (C) “Perspectives on Genetic Counseling” reflect the majority’s belief that genetic counseling is beneficial for individuals to gain knowledge and learn about available options. However, discussions also illuminated concerns about its access and exposure. Responses also suggested that reproductive risk information delivered in genetic counseling can feel impersonal and/or threatening to patients, providing implications for clinical practice.

Section (D) Perspectives on Racialized Views of sickle cell disease and on an Ancestral Framing. Participants perceived negative implications of racialized views of SCD, in the clinical, research, and public contexts. The ancestral framing of sickle cell disease received mixed responses.

Thesis Readers:
Debra Roter, DrPH
Lori Erby, PhD, ScM, CGC
ACKNOWLEDGEMENTS

I would firstly like to thank my parents, two of the most courageous, hardworking, and loving people I know. Only through their sacrifices and risk-taking do I have the freedom to pursue what I believe in and the ability to never give up until I achieve it.

... The completion of this study would have been impossible without the collaboration and collective support of the individuals and institutions/organizations below. To all of you, it has been an honor working and learning together. I am eternally grateful for your help.

My study participants:

Your wisdom, resilience, and altruism are inspiring. It has been an absolute privilege to partner with you for this study, to be part of discussions that were so informative, moving, and brilliantly comedic at the same time. Your insights were compelling and important. As healthcare providers and researchers, we have much to learn from you. Thank you for trusting me with your stories.

National Human Genome Research Institute (NHGRI) and Johns Hopkins School of Public Health (JHSPH):

I thank the NHGRI Intramural Research Training Program for study funding and administrative support. I thank Dr. Debra Roter for serving as my PI, for optimizing this study with your expertise and brilliant ideas, while at the same time providing me autonomy over ideas. Thank you to Dr. Lori Erby for her compassionate support; her wisdom and dedication as a genetic counselor and leader will continue to grow and enhance this field. I am so fortunate to have been trained by Dr. Susan Hannum, an incredibly knowledgeable and skilled qualitative researcher in addition to a truly compassionate mentor. I thank her for the tremendous time you have devoted to me and this study. I thank Mr. Vence Bonham for taking me under his wing, connecting me to resources, and providing guidance through every challenge I faced throughout this process. And, I thank my classmates--Alexis, Hannah, and Laynie--for being there through highs and lows.

Johns Hopkins Sickle Cell Center for Adults:

The Johns Hopkins Sickle Cell Center for being an integral partner throughout this entire. I especially thank Dr. Sophie Lanzkron, Dr. Lydia Pecker, Ms. Natalie Photiadis, Ms. Nikia
Vaughan who have contributed significantly to study development and/or participant recruitment.

**Maryland Sickle Cell Disease Association (MSCDA):**

I thank the MSCDA for their collaboration, with special gratitude to MSCDA President Mr. Derek Robertson who has been incredibly supportive and enthusiastic about this effort. I also thank Ms. Petronella Barrow who has devoted a substantial amount of her time with recruitment.

**Howard University Hospital:**

My many thanks to Ms. Barbara Harrison who has provided great insights towards study development and on data interpretation through the lens of genetic counselor with sickle cell expertise. I also thank Ms. Cynthia Gipson for helping spread the word about this study.

**National Heart, Lung, and Blood Institute (NHLBI):**

I thank the NHLBI Sickle Cell Program for their assistance with participant recruitment, with special gratitude to Ms. Ingrid Frey and Mr. Jim Nichols for generously contributing much of their time towards this endeavor.

... 

I would additionally like to thank the following individuals who have generously contributed their time to study development, participant recruitment, administrative support, and/or document revision:

Ms. Ashley Buscetta, Ms. Khadijah Abdullah, Dr. Sean Bediako, Dr. Andrew Campbell, Ms. Macy Early, Ms. Lauren Claus, Ms. Manuela Montana, Ms. Jessica Gunn, Ms. Chrystal Okonta
# TABLE OF CONTENTS

**ABSTRACT**  

**ACKNOWLEDGEMENTS**  

**TABLE OF CONTENTS**  

**LIST OF TABLES**  

**BACKGROUND**  

**STUDY AIM AND OBJECTIVES**  

**METHODS**  

  - **Sampling and Recruitment**  
  - **Focus Group Procedures**  
  - **Data Analysis**  

**RESULTS**  

  - Participant Characteristics  
  - Focus Group Findings  

  **A. Experiences in the Personal, Social, and Healthcare Contexts**  
    - A1.0 Sickle Cell Disease is More than a Blood Disease  
    - A2.0 Perceived Gap in Empathy for Individuals with Sickle Cell Disease  
    - A3.0 Individuals Seek Partnership with Healthcare Professionals  
    - A4.0 “People Don’t Talk about Sickle Cell:” Low Awareness in the U.S.  

  **B. Dimensions of Decision-Making**  
    - B1.0 Individuals Have to Weigh Multiple Risks and Uncertainties  
    - B2.0 Loss of What Could Have Been  
    - B3.0 “I’m a Mother in a Way, Too”: Individuals Pursue Parenthood through Multiple Avenues  

  **C. Experiences and Perspectives on Genetic Counseling**  
    - C1.0 Genetic Counseling is a Way to Gain Knowledge and Learn Options  
    - C2.0 Genetic Education Can Help Correct Misinformation  
    - C3.0 Barriers to Genetic Counseling: Areas to Address  
    - C4.0 Information May be Perceived as Harmful: Manner of Delivery Matters  

  **D. Views on Racialization of Sickle Cell Disease and on Ancestral Framing**  
    - D1.0 Pigeon-holing of Sickle Cell Disease Can Have Ramifications  
    - D2.0 Ancestral Frame has Potential Significance  
    - D3.0 Relevance of Slavery and Colonialism with Ancestral Frame
D5.0 Ancestral Frame is Not Relevant: Malaria is Still Prevalent and is Harmful to those with Sickle Cell Disease 74

DISCUSSION 76
Views on Awareness and Perceptions of Sickle Cell Disease 77
Healthcare Interactions and Relationships 82
Educational and Psychosocial Dimensions of Reproductive Decision-Making 83
Perspectives on Genetic Counseling 84
Strengths of Focus Groups 87
Study Limitations 88

APPENDICES 91
Appendix A: Recruitment Scripts 91
Appendix B: Verbal Consent Scripts 108
Appendix C: Contact and Availability Form 111
Appendix D: Demographics and Survey Questionnaire 112
Appendix E: Focus Group Facilitator Guide 113

REFERENCES 118

CURRICULUM VITAE 123
LIST OF TABLES

Table 1 Demographic and characteristics of study participants........................................26
BACKGROUND

Sickle Cell Disease and Trait

Sickle cell disease (SCD) has its roots in genetic variants that cause a single amino acid change of beta-globin chain of hemoglobin A. All individuals have two copies of the gene encoding the beta-globin chain. Having one altered copy of this gene confers protection against malarial infection. Those who have one copy of the beta-globin mutation have sickle cell trait (SCT), also known as carrier status for sickle cell disease, and generally, do not have serious health complications (Bender et al., 2016). However, those who possess two copies of the variant described above—inheriting one copy from each carrier parent—have sickle cell disease. An individual who has SCT or SCD has a chance of passing on sickle cell disease to the next generation, depending on the genetic status of his or her partner. For example, in a couple who both have SCT, each pregnancy has a 25% chance of having sickle cell disease, ½ chance of having SCT, and 25% chance of having neither SCT nor SCD. In a couple in which one individual has sickle cell anemia (SS genotype) and the other individual is a carrier for SCD, the probability of recurrence in pregnancy is 50%.

Currently, millions of people worldwide have SCD (Centers for Disease Control and Prevention (CDC), 2010; Hassell, 2010). In the United States, up to 100,000 people have SCD, with the vast majority being African-American or Black. It is estimated that 1 to 3 million Americans have sickle cell trait.

Sickle cell disease is a term encompassing multiple forms of blood disorders with distinct genotypes. The major forms of SCD are sickle cell anemia (hemoglobin SS; most
common), sickle-hemoglobin C disease (hemoglobin SC), sickle beta zero thalassemia, and sickle beta plus thalassemia. Hemoglobin SS and sickle beta zero thalassemia are the most severe forms of SCD and are also referred to as sickle cell anemia. Comparatively, hemoglobin SC is considered moderate in severity and sickle beta plus thalassemia is mildest (Rees et al., 2010).

Patient Experiences: Physical Challenges and Barriers to Care

SCD affects every system in the body and requires lifelong comprehensive medical care. Unfortunately, there is less access to SCD care compared to other common, yet less frequent, hereditary conditions such as cystic fibrosis and hemophilia (Grosse et al., 2009). For cystic fibrosis and hemophilia, national networks of specialty clinics are widely available and reach a larger proportion of target patient populations. Such centers for SCD are both fewer in number, less likely to be networked, and reach a smaller target population (Grosse et al., 2009). Barriers to high-quality sickle cell care negatively affect quality and quantity of life for individuals affected. A study by Lanzkron and colleagues (2013) demonstrated from the National Center for Health Statistics multiple-cause-of-death data (N=16,654 SCD deaths) a significant overall increase in mortality rate of 0.7% among adults with SCD each year during the period of 1975-2005 (Lanzkron et al., 2013).

In sickle cell disease, the amino acid differences in the beta-globin chain of hemoglobin A increase susceptibility of red blood cells becoming rigid, sickle-like structures. Several complications can occur as a result: anemia, painful episodes, long-term pain, infection, stroke, leg ulcers, stroke, and organ failure (Bender et al., 2016). If the condition is left untreated, the risks for complications as well as death increase.
Symptoms and severity of complications are highly variable between individuals, even for those who have the same form of sickle cell disease (Bender et al., 2016).

Historically, there have been few targeted treatments developed for sickle cell disease. Care of people with sickle cell disease includes prevention with vaccination and antibiotics, high fluid intake, folic acid supplementation, and pain medication. It may also include blood transfusion and hydroxyurea medication. Only a small percentage of people with SCD can be cured by bone marrow transplant (Bender et al., 2016). More recently, there have been encouraging advancements on targeted treatments. Progress has also been speeding toward the goal of gene therapy that involves genetically modifying hematopoietic stem cells. Genetic strategies involve using viral vectors containing RNA to target \textit{BCL11A} to increase fetal hemoglobin production or replacing the mutated \textit{HBB} copy with a healthy copy (Demirci et al., 2018).

Despite the genetic simplicity of SCD, there are wide differences in clinical presentations of the disease between affected individuals of the same type of SCD. While some patients live up to 60 years with infrequent crises, others die during childhood or adolescence (Fadare, 2009). Despite advances made in understanding the pathology and treatment of sickle cell disease, predicting the clinical outcome of sickle cell disease is still difficult. Because healthcare professionals are currently unable to predict the clinical course of an affected newborn or fetus to inform parents and couples, newborn and prenatal genetic counseling for sickle cell disease has been a challenge (Fadare, 2009; De Montalembert M et al. 1996; Thein, 2013).

\textit{Patient Social Experiences}
In addition to the physical complications associated with the condition, there is health-related stigma attached to sickle cell disease that contributes to the psychological burden. Stigma occurs when negative stereotypes about a group are applied to a member of that group. In the context of SCD, stigma can be compounded by racism (or racism perceived by the patient) and pose significant barriers to care. Cross-sectional quantitative and qualitative studies have focused on patients' anecdotal reports that seeking specific medication for SCD-related acute pain is often perceived by healthcare professionals as drug-seeking behavior, which translates to poor quality and timeliness of care (Labrousse, 2007; Mathur et al., 2016, Mulchan et al., 2016; Bulgin et al, 2018). These reports have been confirmed by Haywood and colleagues (2013) who performed a cross-sectional, comparative analysis of 2003-2008 data from the National Hospital Ambulatory Medical Care Survey. Findings show that patients with SCD who seek care at the emergency department experience longer wait times relative to other groups, even after accounting for triage level assigned. SCD patients' African-American race may contribute to longer wait times (Haywood et al., 2013).

Stigma related to SCD can also stem from an affected individual’s community. For instance, the extreme pain and fatigue often associated with sickle cell disease may lead to inevitable absenteeism from school and work, creating a false impression that the individual with SCD is “lazy” (Ola et al., 2016; Royal et al., 2011; Dyson et al., 2010; Bulgin et al, 2018). Reports from some studies have mentioned both SCD stigma within the African-American community and lack of desire to discuss sickle cell disease among African Americans (Burnes, 2008; Mayo-Gamble, 2019).
A systematic review of twenty-seven quantitative reports on levels of stigma and qualitative studies that suggest relationship between stigma and well-being and one intervention study on SCD stigma by Bulgin et al. (2018) demonstrates that stigma can hinder physiological and psychological wellbeing and impair healthcare interactions. Several qualitative and quantitative studies have suggested that individuals may feel discouraged to seek necessary professional care if they predict that they will experience stigmatizing behavior from clinicians and may then resort to maladaptive strategies to cope with pain (Jenerette et al., 2014; Labore et al., 2017; Bulgin et al., 2018).

**Race, Ancestry, and Sickle Cell Disease**

Although sickle cell disease is observed more frequently in populations of African descent in the U.S., it is also common among Hispanic people and those in northwestern India and areas around the Mediterranean. Historians and anthropologists pose that the misconception about sickle cell disease as a “Black disease” is still relevant today (Tapper, 1999; Wailoo, 2001; Bediako and Haywood, 2009). Although this topic has been explored candidly by anthropologists and historians, healthcare researchers have less frequently incorporated dialogue about race into a discourse about the social context of sickle cell disease.

In a preliminary online survey study of racially and ethnically diverse adults (60% female; 54% African-American, 28% White, and 18% Asian-American), participants were asked to respond with what comes to mind when they think of ‘sickle cell disease.’ 65/210 of the participants answered with words pertaining to race (e.g. ‘a genetic disease that affects Blacks’; ‘a disease of blacks’; ‘blood disorder common among people of African descent). Of the participants whose responses included a racial perception, all
referred to Blacks or Africa (Bediako and Moffit, 2010). Participants who described sickle cell disease in racial terms also endorsed more intense negative ratings of a hypothetical typical person with sickle cell disease compared to those whose responses did not include a racial perception (Bediako and Moffit, 2010). Although these findings are speculative, they suggest that racial biases tied to sickle cell disease can contribute to or exacerbate discrimination of individuals who have sickle cell (Bediako and Moffit, 2010).

Moreover, findings from Sankar et al. (2006) showed that respondents with sickle cell disease associated their condition with racial identity and discrimination while individuals with cystic fibrosis rarely associated their disease with race (Sankar et al., 2006).

As another example, in a study by Gallo et al., 2013, a Hispanic female participant with SCT and a child with SCD expressed the importance of providing education to other ethnic and racial populations. She reported that she was informed by her provider that SCD was only an “African American disease” (Gallo et al., 2013). Rotimi (2004) argues that due to the erroneous conception that SCD is a ‘black disease,’ most people, including physicians, are not aware that sickle cell disease occurs in the Orhomenos town in central Greece at a rate that is twice that among African Americans or that black South Africans do not carry the sickle cell trait (Braun, 2002; Kevles, 1995). These misconceptions have the potential to lead to negative consequences. For example, a child with sickle cell disease was not correctly diagnosed and almost had unnecessary surgery because he appeared to be of European descent. The child’s parents were of Indian, northern European, and Mediterranean ancestry (Rotimi 2004).
Geneticists, therefore, support that ‘ancestry’ is a better indicator than ‘race’ or ‘ethnicity’ in predicting risk for sickle cell trait and other disease variants (Rotimi, 2004; Fujimura and Rajagopalan, 2011; Ali-Khan, et al., 2011). It has long been proposed that there are five haplotypes of the sickle cell variant, meaning that the genetic alteration occurred five different times throughout history. However, recent evidence has suggested that the sickle cell variant occurred in one individual as long as 7,300 years ago when the world had a population fewer than 5,000 people. Because of its protective benefits, the variant continues to exist today (Shriner, 2018). As humans evolved and migrated across the world, the variant survived among populations who inhabited regions with high malaria rates (Shriner and Rotimi, 2018) This provides an explanation as to why sickle cell disease is more frequent among specific ethnic groups but can occur in any population. Overall, however, there has been a lack of research related to how individuals with sickle cell disease or sickle cell trait perceive the ancestral versus racial nature of their disease or how alternative frames may change perceptions.

**Sickle Cell Disease and Genetic Testing in the US: Historical Perspective**

Due to the hereditary nature of sickle cell disease, genetic testing and genetic counseling are relevant to populations of individuals with both sickle cell trait and sickle cell disease. Genetic testing for sickle cell disease has been available since the 1950s and is relatively efficient and inexpensive. Unfortunately, testing for SCD and SCT throughout the 1970s in the United States is tied to a history of genetic and racial discrimination (Fulda et al., 2006; Markel, 1997). For example, in some states, genetic screening for sickle cell trait and disease was mandatory for African Americans but not for other at-risk groups, perpetuating the misconception that sickle cell disease was solely a “Black
disease” (Markel, 1997). In addressing the National Sickle Cell Anemia Control Act of 1972, President Nixon once noted that the disease was especially pernicious because it “strikes only blacks and no one else” (Naik and Haywood, 2015).

Many of the state and local programs were based on an inadequate knowledge of the genetics of sickle cell disease and as a consequence many of the laws needlessly stigmatized carriers of the sickle cell trait as well as those with the illness. Perhaps most glaring was the apparent ease with which the diagnosis of a heterozygote "carrier status" of sickle cell anemia was used almost interchangeably with homozygous "disease status." The ostracism of sickle cell carriers, unfortunately, became far more than a theoretical concern for African Americans and public health officials.

State and local programs were often based on inadequate knowledge of the genetics of sickle cell disease. As a result, laws were implemented that lead to stigmatization of both carriers of sickle cell disease and individuals affected with sickle cell disease. Diagnosis of heterozygote carrier status was sometimes used interchangeably with disease status (Markel, 1997). Due to misinformation about SCT versus SCD, many healthy African Americans who were identified to have sickle cell trait were denied educational opportunities, employment, life and health insurance, and entrance into the U.S. armed forces due to confusion about the implications of sickle cell trait (Markel, 1997). The confusion also led to people being inaccurately counseled on their health or children's health (Naik and Haywood, 2015). For example, parents of children who tested positive for sickle cell trait have reported being misinformed that their children would become very ill and die early (Naik and Haywood, 2015).
Current Genetic Testing and Counseling Practices in the Context of Sickle Cell Disease

Today, genetic testing for sickle cell disease and trait occurs largely through newborn screening and through carrier testing in the prenatal setting.

In 2006, all states in the U.S. required and provided universal newborn screening for SCDs and other hemoglobin disorders. Within 2 to 3 weeks after newborn screening, results are sent to the baby’s healthcare provider who is to recommend quick follow-up testing and/or treatment or management as necessary. There is currently wide variability in state policies regarding notification to parents on test results and referrals to genetic counseling (Naik and Haywood, 2015; Taylor et al, 2014). Although all parents are notified of a positive SCD result, only 37% of parents are notified if their child has SCT (Naik and Haywood, 2015). For the parents who receive SCT screening results, it is not guaranteed that they are informed about the hereditary implications of SCT or would remember to share results with the child later to inform reproductive decisions. There are no systematic processes in place to facilitate later communication with the child as an adult. Depending on the location in which children receive their care, parents may receive genetic counseling on sickle cell trait, either with a genetic counselor or a pediatric provider. Other times, this information is transmitted in the mail, which may get lost over time. Furthermore, if the child does not maintain the same physician or medical network they had at birth, test results could be lost as the child transitions through multiple medical systems (Naik and Haywood, 2015).

In the prenatal setting, the American College of Obstetricians and Gynecologists (ACOG) recommends screening for carrier status of hemoglobin disorders among
pregnant patients based on their ethnicity. ACOG recommends that hemoglobin testing occurs among patients of African, Mediterranean, Middle Eastern, Southeast Asian, and West Indian descent (ACOG Committee on Obstetrics (2007)). For a couple who are both carriers of the same disorder, they are offered prenatal testing of the pregnancy to inform a deeply personal decision on continuing the pregnancy or terminating (Pecker et al., 2018). Alternatively, preimplantation genetic diagnosis after in vitro fertilization is a procedure that allows for implantation of unaffected embryos. This procedure, however, is expensive and typically not fully covered by health insurance, even for patients who are at risk of transmitting a genetic condition (Pecker et al., 2018).

In general, reproductive decisions are deeply personal and influenced by a complex interaction of personal, ethical, cultural, and religious values as well as socioeconomic circumstances. For parents at risk of conceiving a pregnancy with sickle cell disease, a study by De Montalembert M et al. (1996) has shown that cultural reasons, religious reasons, educational level and the number of children in the family play roles in the decision to request a prenatal diagnosis.

Genetic testing is sometimes accompanied by genetic counseling with a genetic counselor. Genetic counselors are health professionals who, along with providing the patient necessary genetic information, facilitate decision making that is consistent with a patient’s social, ethical, and cultural values (Resta et al., 2006). As opposed to gaining information through educational materials, interpersonal interaction with a genetic counselor allows for a conversation that is tailored to the individual’s values and preferences. It also allows patients to address related psychological and social issues and to directly ask questions to clarify understanding of the information they are provided.
Genetic counseling may be cost-prohibitive to some patients, as there is currently variability in cost coverage of genetic counseling and testing among private health insurance companies and among state Medicaid programs (Andrews et al., 1994). The percentages of couples “at-risk” and parents of affected newborns who are referred to genetic counseling are not documented to our knowledge. Health professionals advocate for an increase in access to genetic counseling for the sickle cell disease and sickle cell trait populations (Taylor et al., 2014).

**Literature on Genetic Counseling in the Context of Sickle Cell Disease**

Despite health professional support for increased access to genetic counseling related to SCT and SCD, there is limited research on genetic counseling in the context of sickle cell disease.

*Communicating Sickle Cell Disease to Patients in Genetic Counseling*

As previously mentioned, sickle cell disease is highly variable in its clinical manifestation and severity, and there are no early prognostic factors. Consequently, genetic counseling related to sickle cell disease in both the prenatal and postnatal contexts has been considered difficult medically and ethically because it is not yet possible to predict and therefore communicate the severity disease in an unborn child during prenatal diagnosis (de Montalembert M et al., 1996). There are guidelines from the National Society of Genetic Counselors that specify best practices on communicating a prenatal or postnatal diagnosis of variable conditions like Down Syndrome (Sheets et al., 2011). To our knowledge, no such similar genetic counseling guidelines yet exist for sickle cell disease. Studies that could contribute to the development of genetic counseling
communication guidelines for sickle cell disease would have potential value for both clinician and patient stakeholder groups.

Lay Conceptions of Sickle Cell Trait and Disease

Several studies have explored lay conceptions of sickle cell trait and disease among individuals of reproductive age from the general public, most often African-American individuals (Treadwell et al., 2006; Gustafson et al., 2010; Williams-Smith, 2015). Focus group and survey studies suggest that the majority of people with known SCT status were more aware of the inheritance pattern and reproductive significance of SCT compared to people with unknown status (Treadwell et al., 2006 and Mayo-Gamble et al., 2018). However, while some participants in this focus group study understood the reproductive implications of SCT, this knowledge was limited. For example, some participants reported that they do not have a complete understanding of sickle cell disease or the probability a future child would inherit the condition (Mayo-Gamble et al., 2018). Similarly, in a study by Long et al. (2011), participants (including those aware of SCT status) understood the natural progression of sickle cell disease but had limited understanding of its inheritance. Furthermore, a quantitative study by Gustafson et al. (2010) surveying African-American women (N=101) concluded that African-American women have a relatively high belief in the severity of sickle cell disease but often do not believe they are at risk of having a child with SCD (Gustafson et al., 2010). There is limited research exploring appropriate ways to deliver information to communities.

Unmet Information Needs

Some studies have looked at the unmet informational needs of people with SCT and people who have increased chances of having SCT (Housten et al., 2016; Mayo-
Gamble et al., 2019; Pass et al., 2010). For example, a focus group study by Mayo-Gamble et al. (2019) revealed that people with SCT reported not being given education about SCT and SCD by their healthcare providers, having informational resources that inadequately conveyed the reproductive implications of SCT, and receiving unclear communication about SCT and SCD in the interpersonal, community, and medical settings. Participants from this study also argued for the importance of universal sickle cell screening across populations, not just among African Americans (Mayo-Gamble et al., 2019). While there may be other systematic ways to improve awareness, medical experts argue that lack of a universal method of family notification among newborn screening programs and variation in policies and practices regarding disclosure of sickle cell trait are responsible for low awareness of SCT status in the United States (Gustafson et al., 2007; Pass et al., 2010).

Furthermore, a few studies have also researched the effectiveness of genetics tools for young adults with SCT and SCD, examining the effectiveness of tools assessing users’ knowledge about SCD and SCT and providing education to address the gaps in knowledge described above. A study by Wilkie and colleagues (2013) set out to improve knowledge, intention, and behavior related to reproductive health among individuals with SCD and SCT through a web-based, multimedia intervention. Findings show that the intervention group receiving the training tool had significantly higher average knowledge scores and probability of reporting a parenting plan to avoid SCD or SCD and SCT in the next generation compared to the group that was provided an e-Book. The intervention group also showed significant change in intention and planned behavior. The authors conclude that findings provide the groundwork for testing the effectiveness
of the tools on a national level (Wilkie et al., 2013; Gallo et al., 2016). These tools are computer-based and do not directly involve genetic counseling through a clinician. The experience of genetic counseling with a clinician involves providing personal genetic risk information, facilitating decision making based on clients’ preferences and values, and often providing psychosocial support. However, further research is necessary to examine how best to provide genetics services to individuals with SCD and SCT.

Reproductive Attitudes and Behaviors

Some studies have explored reproductive attitudes and behaviors among people with SCT, SCD, and members of “high-risk” racial groups. Many of these studies have been conducted with people outside of the United States, for example in West Africa, where rates are relatively very high (Adejumo and Olaoye, 2018). Smith and Aguirre (2012) examined reproductive attitudes and behaviors in people with SCD or SCT through a qualitative interpretative meta-synthesis of three studies on this topic (1 UK study and 2 US studies).

While participants expressed diverse attitudes and behaviors, common themes emerged from the synthesis: lack of awareness across groups; denial of having SCT among some male participants, unexpectedness in learning one’s SCT status; value of relationships over SCD risk; and challenges of asking about partners’ SCT status and in making difficult prenatal and abortion decisions. The synthesis concludes that education and awareness play a role in reproductive attitudes and behaviors (Smith and Aguirre, 2012).
A focus group study (N=15) in the U.S. by Gallo et al. (2010) offers an in-depth exploration of the factors and barriers that influence reproductive decisions among participants with sickle cell trait and sickle cell disease. Among these participants, reproductive decisions were influenced by the desire to have children and the increasing availability of treatments that improve quality of life and longevity (Gallo et al., 2010; Smith and Aguirre, 2012). These decisions were also influenced by religious and cultural values for some, but not for others (Gallo et al., 2010). The participants with SCD noted that their own health problems also presented struggles in family planning. Specifically, they noted that women with SCD have difficult pregnancies and fear that a shortened lifespan would prevent them from caring for their children. Furthermore, most participants in this study expressed the importance of people knowing their genetic status and reproductive risk and sharing SCT status with partners; participants voiced concern about young people lacking knowledge and understanding about sickle cell inheritance (Gallo et al., 2010).

Therefore, studies in the extant literature have focused on the unmet informational need for genetics education and the challenges and barriers related to reproductive risks for members of the SCD and SCT populations. These studies highlight the need for research to better understand strategies to address these unmet needs and barriers.

**Racial Differences/Disparities in Genetic Testing Uptake**

Outside the sickle cell context, a number of studies have shown differences in clinical genetic testing uptake among African-American/Black individuals and White individuals. In the prenatal context, Kupperman and collages (2016) showed in a review
and assessment of medical charts of 238 women 35+ years seen by 20-weeks gestation from the University of California, San Francisco that Latinas and African-American women were much less likely to undergo prenatal diagnosis than were whites and Asians. With women as the reference group, OR and CI, of Latinas were 0.28 (0.09-0.83) and African Americans were 0.33 (0.10-1.10), after adjustment of socioeconomic characteristics (Kuppermann et al., 1996).

Studies suggested that these disparities may be explained by a combination of attitudes towards genetic testing and barriers to accessing genetics services. For example, a cross-sectional survey with 430 (170 African-American and 181 Caucasian) respondents showed that awareness of predictive genetic testing was higher among Caucasians than African-Americans. African Americans were also more likely to believe genetic tests would be used by the government to label certain groups as inferior and less likely to endorse health benefits from genetic testing (Peters et al., 2004). Another analysis of a nationally representative sample of 1,724 adult men and women demonstrated racial differences in knowledge and concerns regarding potential misuse of genetic testing. There were differences in health insurance coverage by race/ethnicity as well as significantly higher levels of mistrust in a physician by a medical system (Suther and Kiros, 2009). Lastly, McCarthy and colleagues (2016) conducted a population-based study including adult women in Pennsylvania and Florida who were diagnosed with breast cancer between 2007 and 2009. The study included cancer registry data, AMA Physician Masterfile, and patient and physician surveys of 3,016 women, medical oncologists, and surgeons. Black women were significantly less likely than white women to undergo BRCA1/2 testing, after adjusting for mutation risk, clinical
factors, sociodemographic characteristics, and attitudes about genetic testing. Adjusting for clustering within physician or physician characteristics did not change size in testing disparity, suggesting that racial differences in testing are also largely attributable to differences in physician recommendations.

Because of a history of racially-focused eugenics and historical experiences like the Tuskegee experiment in the United States, it is important to understand and address differences in attitudes towards genetic testing. Furthermore, also important is the need to address barriers to genetics services that may be more prevalent in racial minority groups.

STUDY RATIONALE

Medical professionals express support for increasing access to genetic counseling in the context of sickle cell disease. However, there is still a gap in knowledge about patients’ experiences, perspectives, and insights pertaining to genetic counseling with a health professional. We argue that in order to advocate for an increase in access to clinical genetic counseling related to sickle cell disease, it is essential to improve our understanding of ways to enhance the benefits and mitigate any perceived harms of genetic counseling for stakeholder patient groups. Despite the relevance of genetic counseling and testing for the SCD and SCT populations, the history of genetic testing in the sickle cell disease context in the U.S. may discourage seeking help from a clinician.

Additional information would be important for clinicians who wish to deliver valuable, culturally-sensitive genetic counseling. Our proposed study would add to the literature by exploring areas for improvement in genetic counseling. The immediate purpose of this study is not to draw generalized conclusions about the experiences and
perceptions of the members of these populations. There is high diversity within both the SCD and SCT populations, and genetic counseling preferences are likely heterogeneous within both groups. Instead, this research aims to explore common experiences and insights of individuals from the sickle cell disease population that could assist with the tailoring and personalizing of genetic counseling and highlight areas for further research.

**STUDY AIM AND OBJECTIVES**

The overall aim of this study is to gain a better understanding to enhance the benefits and mitigate the perceived harms of genetic counseling in the sickle cell context. The following objectives facilitate a better understanding of experiences of and perspectives on genetic counseling as well as a broader understanding of the healthcare experiences of individuals with sickle cell disease.

*Objective 1: Explore participants’ experiences of living with sickle cell disease.* To explore the range of ways that participants describe sickle cell disease and the ways that sickle cell disease affects the individual and the family, including discussions of the complexity/variability of the condition, how the experience is bounded by culture, and any experiences related to societal reactions or stigma.

*Objective 2: Explore experiences around genetic counseling with clinicians (or genetic education from non-clinicians) and understand perceived benefits and or harms from the experiences, as perceived by individuals with sickle cell disease.*

*Objective 3: Explore perceptions of racialized beliefs in the context of sickle cell disease and to explore reactions to an ancestral framing to describe...*
sickle cell diseases protective to survival. To explore how participants frame the underlying cause of sickle cell disease and their perceptions of the way(s) in which others view the condition’s cause. More specifically, to capture participants’ thoughts on a commonly cited societal belief that sickle cell disease is a “Black disease” versus an “ancestral disease” and to assess the extent to which participants perceive these beliefs exist in the medical community. To explore reactions to both kinds of framing.

METHODS

A qualitative descriptive approach was used to meet the objectives of this study. Descriptive studies are often important when investigating social experiences and phenomena that are unknown, distinctive between subjects of a target population, or have become stereotyped (Rowles & Reinharz, 1988; Hammarberg et al., 2016). These characteristics reflect the nature of our study questions. The topics of investigation, as they pertain to the sickle cell population, have been underexplored in both qualitative and quantitative social science literature. Secondly, we expected responses to be diverse and specific to the individual participants’ views and experiences and therefore best captured through a qualitative approach. A qualitative approach allowed participants to share their experiences and perspectives in their own words.

Because of the nature of both the research question and the study population, the method of qualitative focus groups was chosen. The ultimate goal of the research is to incorporate the opinions of people with SCD into genetic counseling practice. Focus group methodology was chosen to allow participants the opportunity to speak to each
other’s experiences and agree/contend with each other’s perspectives. Although focus
groups are logistically more complicated and allow less time for each participant to
contribute to the overall data relative to other qualitative inquiry methods, they allow
access to insights that result from the interaction between the participants (Morgan,
1988). Furthermore, all participants belonged to minority ethnic/racial groups given the
geographic area of recruitment. In a randomized controlled trial with African-American
men (N = 350), Guest et al. (2017) demonstrated that several types of sensitive and
personal themes (on healthcare- and health-related topics) emerged in the focus group
setting that did not emerge in the individual interview setting. Finally, we selected the
focus group methodology with the goal that a higher participant to researcher ratio
would enhance comfort in the participation experience and provide participants with
more autonomy over the content of the discussion.

This study was determined as exempt (category 2) by the Johns Hopkins
Bloomberg School of Public Health and the National Institutes of Health Institutional
Review Boards.

**Sampling and Recruitment**

Adult individuals were recruited through convenience sampling. Individuals were
eligible if they had a diagnosis of sickle cell disease (any genotype), were 18 years of age
or older, and were able to speak fluent English. Convenience sampling is a type of non-
probability sampling in which participants are recruited based on accessibility and
proximity to the researcher. This method facilitated recruitment for in-person focus
groups. To a lesser degree, snowball sampling also occurred as some participants had shared the study with other eligible individuals in their networks.

Recruitment was focused primarily in the Baltimore and Washington D.C. area in the Mid-Atlantic region of the United States, occurring at the following sites: Johns Hopkins Sickle Cell Center for Adults, Maryland Sickle Cell Disease Association (MSCDA), National Human Genome Research Institute (NHGRI), National Heart, Lung, and Blood Institute (NHLBI), Howard University Hospital, and the Sickle Cell Disease Association of America (SCDAA). Recruitment occurred through both active and passive means. Passive recruitment involved flyers being made available in areas such as sickle cell disease clinics in order for eligible individuals to self-refer by calling the researcher. At the Maryland sickle Cell Disease Association, an advocacy group leader distributed flyers to group members. Active recruitment involved an associate investigator clinician asking for permission of eligible individuals seen in clinic to be contacted by the lead researcher or to be introduced to the lead researcher for invitation to the study.

The researcher conducted five focus groups with the attempt to recruit five to seven participants per group. Focus groups were coordinated based on participant availability. To help reduce the impact of no shows, the researcher attempted to over-recruit by 2-3 participants for each focus group discussion.

**Focus Group Procedures**

After individuals were recruited, they entered the consent phase of enrollment. Potential participants either called the lead researcher or had given their permission and contact information to be contacted by DP to learn more about the study. Over the phone,
DP provided more details about the study. If the individual expressed interest in proceeding, DP and the individual discussed enrollment and consent information per recruitment script (Appendixes A & B). If the individual provided consent, DP asked the individual for general availability, dates and preferred location to meet for study participation. DP organized focus group dates based on availability of the most number of individuals. DP re-contacted individuals who had not been able to meet on available dates to attempt to include them in future focus groups.

Three focus group discussions were conducted at the Johns Hopkins Medical Institute campus, one discussion group occurred at the National Institutes of Health, and one discussion group (Group 5) occurred over WebEx conference call. Verbal informed consent was obtained with each participant again among the group prior to each focus group discussion. Individuals who no longer wished to participate had the opportunity to leave the study, but none who attended the group consent ultimately chose to leave.

During each focus group, the lead researcher and focus group facilitator (DP) posed open-ended questions from a focus group guide developed for this study and intended to address the three primary objectives. Each focus group was scheduled for 1.5 hours, but some discussions extended to 2 hours. DP took notes during the focus groups that included participants’ non-verbal expressions and responses (e.g. nodding in agreement) and confusion with certain questions.

Minor modifications included changing the wording of the questions to increase clarity in the moment, altering the order of questions to improve discussion flow, and wording of the questions. Additionally, during the discussions, the facilitator asked open-ended questions related to responses participants provided. These questions were asked
for further clarification or elaboration on responses that were related to questions or probes on the question guide. Participants completed a questionnaire (Appendix D) that included questions about demographic information and specific aspects of experiences with receiving genetic counseling services, either before or after the focus group discussion, depending on when they showed up.

At the closure of the focus groups, each individual was provided a $50-value gift card for their participation. Parking validation and light refreshments were also provided.

**Data Analysis**

Analysis of data was founded on inductive thematic analysis constructed within a critical realism paradigm. Analysis processes were informed by application guidelines defined in Braun and Clarke (2006). A critical realism approach was determined to be appropriate because our goal was to evaluate data and suggest appropriate changes to identified problems accordingly. Further, the discussion questions were developed with the assumption that there is a relatively straightforward relationship between meaning and experiences and perspectives, which makes analysis of the data compatible with a realist paradigm.

The audio recordings were transcribed by a professional transcription company. The lead researcher/facilitator (DP) gained familiarity with each transcript and field notes that were collected during the focus groups by doing an initial read of all documents, while writing memos about observations. DP collaborated with two assistant investigators to discuss preconceived ideas and identify repeated patterns and ideas
from one initial transcript. Excerpts and segments of data were labeled with codes, which are defined by Boyatzis (1998) as the most basic segment, or element, of the raw data or information that can be assessed in a meaningful way. Initial codes were produced by DP from the raw data and derived from the moderator guide. Coding proceeded in an iterative fashion; as new codes emerged as analysis of transcripts progressed, transcripts were re-coded to reflect new codes that emerged. DP independently carried out coding manually on the paper transcripts, and the codes were ultimately transferred to MAXQDA software system (2018, Release 18.2.0) to facilitate further sorting and analysis. Codes were sorted and collated into potential overarching themes. Based on the iterative coding and thematic analysis, DP summarized the data. The PI, who read all five transcripts, reviewed the summary and posed questions. DP returned to the data as necessary to clarify the data presentation while ensuring that the description captured the meaning expressed by the participants. The interpretation involved efforts to theorize the data into themes that capture broader meanings and implications of the summarized data (Patton, 1990). The themes were subsequently defined and titled to reflect the overarching essences of the data. Largely because of the paradigm selected, the themes in this study were identified exclusively at the semantic (descriptive) level, meaning they were identified within the explicit or surface meaning of the data.
RESULTS

Participant Characteristics

Five focus groups were conducted with 21 adult participants who have sickle cell disease, divided into groups of three, four, seven, four, and three participants. Four focus group discussions were held in-person and one group was held over conference call. Thirteen participants were self-identified as female and eight participants as male. All 21 participants identified as Black or African American. The median age range of male participants was 26-45 and the median age range of female participants was 26-35. Median age range of all participants was 36-45. The minimum age range was 18-25 and the maximum age range was 66-75. Six participants reported having children and zero reported having children affected with sickle cell disease. Four reported they had met with a genetic counselor or genetic specialist; three of those four individuals received genetic counseling in the clinical context. Focus group distribution and participants' self-reported demographic characteristics are characterized in Table 1.
Table 1. Demographics and characteristics of study participants.

<table>
<thead>
<tr>
<th>Group 1 (n=3)</th>
<th>Age Range</th>
<th>Race and Ethnicity</th>
<th>Gender</th>
<th>Has children</th>
<th>Met with a genetic counselor</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>26-35</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>B</td>
<td>18-25</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>C</td>
<td>26-35</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>Group 2 (n=4)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>D</td>
<td>18-25</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>E</td>
<td>46-55</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>F</td>
<td>36-45</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>G</td>
<td>26-35</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Group 3 (n=7)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>H</td>
<td>56-65</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>I</td>
<td>66-75</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>J</td>
<td>36-45</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>K</td>
<td>26-35</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>L</td>
<td>26-35</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>M</td>
<td>46-55</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>N</td>
<td>36-45</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>Group 4 (n=4)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>O</td>
<td>36-45</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>P</td>
<td>26-35</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>Q</td>
<td>36-45</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>No</td>
<td>Unsure</td>
</tr>
<tr>
<td>R</td>
<td>46-55</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Group 5 (n=3)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>S</td>
<td>18-25</td>
<td>Black or AA (non-Hispanic)</td>
<td>Male</td>
<td>No</td>
<td>Unsure</td>
</tr>
<tr>
<td>T</td>
<td>18-25</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>U</td>
<td>26-35</td>
<td>Black or AA (non-Hispanic)</td>
<td>Female</td>
<td>No</td>
<td>No</td>
</tr>
</tbody>
</table>

Focus Group Findings

A. Experiences in the Personal, Social, and Healthcare Contexts

A1.0 Sickle Cell Disease is More than a Blood Disease

The experience of sickle cell disease is highly variable among individuals affected. However, a ubiquitous theme that occurred across focus group discussions was the extensive effects the disorder has on the lives of those affected by it. For example, the two participants below describe how the disease has a continuous impact on individuals,
even in the absence of a crisis, and how it requires those affected to be constantly adapting:

“So, before I even discuss the pain, I just let them at least rationally see that, in terms of a day-to-day just waking up, you and I will never be at the same level because I’m half--you know what I mean? I’m running on half of what you’re running on” --Participant K (Group 3), female, 26-35 years old.

“Anyone living life with this disease has become accustomed to having to do everything differently. So, it’s not a normal, standard lifestyle. So, because that has been internalized and that is something that we take into account even just going outside” --Participant R (Group 4), female, 46-55 years old.

Participants expressed that people who are unfamiliar with sickle cell disease seldom fully comprehend its severity. The pain experienced during a crisis is very extreme and some explain that there can be baseline pain, even without the event of a crisis. Participant A describes how he tries to put the experience of a crisis into perspective for those who do not have sickle cell:

“Doctors and physicians who aren’t familiar with sickle cell, I try to explain the way my pain feels as best I can describe it to them. Like, for me, it’s like when you get your hand jammed in a door or you stub your toe really, really hard, but it doesn’t go away. It’s just that permanent throbbing until you get relief from, you know, whatever means they relieve the pain. So, being able to put it in a way where people are like, “Oh, yeah, I know what that pain feels like,” and to say, “it’s that, but it doesn’t go away.” That kind of puts it into perspective as how excruciating it can be and why it’s important to treat it as quickly as possible” --Participant A (Group 1), male, 26-35 years old.
Participant D sheds light on the unpredictability and life-threatening nature of crises:

“I usually, like, highlight the randomness of it sometimes too and, like, how it’s out of the control of, like, okay, like if I get too hot, too stressed, too over-exacerbated, anything like that. If the pressure changes too much or something like that that could trigger a crisis. And I explain, like, what a crisis is, like, when blood’s not getting to a certain part of the body, [...] And then just, like-- And then, like, the thing that usually it’s, like, gets most people is, like, you just have to highlight, like, it can happen anywhere that blood flows. So, like, if it happens in your brain that’s a stroke. If it happens in your chest, like, you know kind of thing” --Participant D (Group 2), male, 18-25 years old.

Participants emphasized that the effects of sickle cell disease do not simply consist of intermittent pain crises and otherwise normal lives. Each crisis causes damage to the individual, both “physiologically and mentally:”

“If it was just the crises, like, okay, I’ve been through that and done. I’m back to normal. No. It’s, like, it now has caused damage with every crisis, it leaves a mark of damage, you know. So it’s, you know” --Participant F (Group 2), female, 36-45 years old.

“Not back to normal, though” --Participant E (Group 2), male, 46-55 years old.

<laughter>

“You’re not. Never back to normal” --Participant F (Group 2), female, 36-45 years old.

“There are consequences” --Participant E (Group 2), male, 46-55 years old.
“You’re a little bit fractured every time” -- Participant E (Group 2), male, 46-55 years old.

Other participants explained that sickle cell disease can be comorbid with other disorders. They also noted that individuals not only experience complications from the disorder directly but can also suffer from side effects of medications they take for management.

A2.0 Perceived Gap in Empathy for Individuals with Sickle Cell Disease

In describing their overall experiences living with sickle cell disease, participants shared their perspectives on stigma-related accounts in both the healthcare and social contexts. Healthcare-related stigma often involves labeling patients as “drug-seekers,” distrusting their reports of pain, and not providing care in a caring manner. A common theme of responses is that stigma was perceived by participants to be associated with apathy towards sickle cell disease, which is exacerbated by othering of the individuals affected with the disorder. The racial minority status of sickle cell disease was explained to greatly contribute to these attitudes. However, based on responses, sickle cell-related stigma may not be explained by race alone. Participants provided examples that suggest that qualities such as relatability and visibility of a disease or disorder may influence the ability of others to empathize with individuals affected. The following subthemes are organized to reflect the multiple contexts this theme encompasses.

A2.1 Patients Prioritized Based on Skin Color, Appearance, and Perceived Socioeconomic Standing

Participants expressed experiencing disproportionately longer time-to-provider wait times in the ER compared to other patients. Across four groups, participants
recounted experiences of seeking emergency services for sickle cell-related pain and observing disparate wait times. One participant shared that he believes this disparity was attributable to how the perceived seriousness is dependent on the race of the individual suffering from the pain.

“This is well before I started going to [HOSPITAL], things like that, to the point where I’m in the ER waiting for three hours while I’m watching other people get called ahead of me. And the part that hurt was, like, you know, looking at the race of people who were called before me. And which is why I think that because sickle cell is a black disease it’s not taken as seriously or people’s pain isn’t taken as seriously if they’re a Person of Color, whether they’re black, whether they’re Hispanic, whether they’re Asian. People of Color’s pain is not taken as seriously as white people in this country. Point blank, period.” --Participant A (Group 1), male, 26-35 years old

In addition to unequal wait times, participants in several groups observed apparent differences in communication and demeanor towards patients based on patients’ race in the emergency department. One participant described how depending on the “hue” of the patient, they would either be treated with impatience or cared for as if they were a loved one.

“I’ve seen where there has been a degree of difference in how they treat certain patients who come through. I’ve seen them take care of patients with certain hues that came in the door and then as soon as a different type of hue came in they were agitated or short-winded. And it was just like, and then when I saw someone else come through it was just like, "Is everything okay?" It was almost like they were talking to, like, their friend or their mother or their aunt.” --Participant F (Group 2), female, 36-45 years old
Lastly, participants across three focus groups reported ensuring that they look presentable to avoid prejudice, emphasizing how they never fail to dress formally when seeking emergency care, regardless of the pain they are suffering.

“No matter how bad my crisis is, when I go into the ER, I don’t dress-- I dress up instead of dress down. Irregardless of how bad I feel. Because the truth is, people judge books by their cover on a daily basis, and because I know my name and my color work against me here-- I hate to say it, but it’s just the truth-- I never look a mess so that it doesn’t give them a reason to assume less about me” --Participant K (Group 3), female, 26-35 years old.

In Group 2, participants discussed how the less ethnic a Black patient is perceived, the greater the chance they will be cared for fairly:

“You might be okay if you’re light-skinned and straighten your hair, you might be” --Participant G (Group 2), female, 26-35 years old

“Yeah, pretty much. Yeah <laughs>” --Participant F (Group 2), female, 36-45 years old.

“Don’t have locks, now” --Participant G (Group 2), female, 26-35 years old

“Okay. <laughs> Let me relax my hair so you could feel relaxed” --Participant F (Group 2), female, 36-45 years old.

Race contributes an obvious role in the stigma of sickle cell disease and responses reflect how participants believe colorism and classism may also be at play. In Group 1, participants discussed that others must realize neither the African-American population nor sickle cell population are not “monolithic.”
“So, there are so many different subcultures within the African-American culture. I mean, you have wealthy individuals with sickle cell, educated individuals with sickle cell, those who are not educated. So, like, again, you can’t throw everyone in the same box” --Participant C (Group 1), female, 26-35 years old.

A2.2. Black Women’s Pain is Not Taken Seriously, Regardless of Sickle Cell Status

Across three focus groups, participants brought up the relevance of neglect of the health and suffering of Black women in the reproductive context, to assert how the pain of African-American or Black individuals is dismissed more broadly. The participants below conveyed that dismissal of pain is evidenced by disproportionately higher maternal death rates in the reproductive setting:

“And even if you don’t have sickle cell, black women in this country, their pain isn’t taken as seriously, because they have higher birth-related death rate than any other minority in this country. Because their pain or their symptoms aren’t taken seriously” --Participant A (Group 1), male, 26-35 years old

They also described the erroneous but pervasive notion that Black individuals are able to tolerate more pain.

“I’ve actually heard, like, people who have gone to nursing school and doctors actually say that, like, and I think it dates all the way back to, like, you know, slavery days, where it’s like black people are thought of to be able to withstand pain more than other races. And so, like, some-- I’ve heard just-- It’s coming out more in the news now, like, just articles about how black women are not really believed as much. Not even with sickle cell, just maybe having a baby” --Participant G (Group 2), female, 26-35 years old

A2.3 Relatability and Visibility of a Disorder affects the Empathy it Engenders
Although race was suggested as the main contributor, participants discussed that disparities in empathy are also rooted in the visibility and/or relatability of the disease. Several participants reported immigrating from countries in West Africa, where they also experienced or observed stigma and discrimination of sickle cell disease. For example, Participant Q from Group 4 reported that he once sought emergency care for a broken leg while living in Nigeria. He noted that even though the pain from his broken leg was not as severe as that of a pain crisis, he received considerably greater attention for his broken leg because the broken leg was visible and a generally more relatable experience than a sickle cell crisis:

“I have had a broken leg once. The pain wasn’t as bad. To a 7 crisis on the 0 to 10 scale. I swear to you, they didn’t care that I did not have a doctor's note. They didn’t care that I missed school for three, four days. They didn’t care about-- They was, they saw the broken leg and they could relate to that and--So here you are, you’re all gravy. But I’m having a 9 crisis and so I can’t show up. And, "Well, there’s something fishy about you. Like, why aren’t you--? Why didn’t you come to-- Why aren’t you in class? "Oh, I had a--" [...] And this is in Nigeria that is black, and it’s worse here in the U.S.”

Participant Q (Group 4), male, 26-35 years old

Another participant described how her parents had concealed her diagnosis of sickle cell disease from her until she was ten years old. She explained that in addition to her parents wanting her to be able to better understand the disease, there was a conscious stigma in Nigeria, where she was living at the time. Before, she recounted her young self bullying a peer living with sickle cell disease and later on realized she was bullying a “mirror” of herself:

“My parents didn’t tell me I had sickle cell until I was ten, and that was because they felt like I wouldn’t understand it. So
they wanted me to get a better understanding of it before--and because in Nigeria it's still a conscious stigma, they didn't want people to segregate me because of it. Now, I'm coming from a place of at seven I bullied a kid who had sickle cell. I was one of the more popular kids, and you know how once you have anything that makes you weird or different, people pick on you” --Participant K (Group 3), female, 26-35 years old.

Participants in Group 2 also had formerly lived in Nigeria and shared experiences living with sickle cell disease there. They described that people in Nigeria generally have a much better understanding of the disorder because it affects a higher proportion of individuals in the country. Although healthcare professionals were knowledgeable and handled sickle cell disease seriously, participants each brought up how sickle cell disease is “looked down upon” or “pitied.” Although people had a better understanding of sickle cell disease, one participant described that individuals in power (e.g. teachers) sometimes do not fully comprehend or empathize with the challenges of those living with sickle cell disease. He suggested a need for individuals in power to have a different perspective on sickle cell disease: “You know how you do not think cancer is that person's fault?”

Further, across all five focus groups, individuals compared the discrepancy between sickle cell disease and other conditions, most frequently cancer, in terms of attention people show to the conditions. Other examples provided by participants reflected the belief that the invisibility of sickle cell disease affects the empathy it engenders, both within the healthcare setting and in the greater community:

“If you come in bald with cancer, nobody's going to say anything to you. It would be never-ending empathy. But, like,
this is invisible, you know” --Participant P (Group 4), male, 26-35 years old

“Like if you see a person with cerebral palsy and they’re there in this wheelchair and they can’t walk and they can’t move. But sometimes, you know, that’s our reality, but you really can’t see it. And so I think the invisible part of the sickle cell is what makes it harder for us. Because we all are sitting here and we just look like, you know…"--Participant F (Group 2), female, 36-45 years old

“Normal people” --Participants D, E, and G (Group 2) in response to Participant F

Participants’ experiences demonstrate that stigmatization of sickle cell disease occurs in countries outside of the United States, countries in which individuals affected by sickle cell disease belong to the racial majority group. Participants suggest that factors that contribute to sickle cell disease stigma are complex and perhaps extend beyond race, perhaps involving aspects such as relatability and visibility of the disease. It is undeniable from the stories shared by participants, however, that race is seen to play an important role in stigma occurring in the U.S.

A3.0 Individuals Seek Partnership with Healthcare Professionals

In the healthcare setting, participants emphasized the necessity of healthcare professionals to both improve knowledge about sickle cell disease as well as work collaboratively with patients. Many individuals share that they try to come to any medical visit, equipped with all the medical information that their providers may request. The following participant described how patients understand that the situation is complex and that sickle cell disease is difficult to treat and manage. She emphasized that the path
to a solution requires healthcare professionals and patients working together collaboratively on the issue:

“I think having that understanding that yes, it is complicated and it’s even complicated for a hematologist who studies this to treat it, I think everybody just understanding that would help everybody realize that it’s complicated, and it’s going to take, you know, a lot of working together” --Participant G (Group 2), female, 26-35 years old.

Similarly, participants like those in Group 1 spoke of the need to acquire adequate knowledge about sickle cell disease among healthcare professionals, but also noted that a trusting relationship between the patient and provider enables them to work together when the information is not available:

“And if you are not educated on the disease, like, go on the Internet and pick up a book. It’s not that hard to educate yourself on any disease nowadays [...] I’m not saying write a dissertation on it, but just research it”--Participant C (Group 1), female, 26-35 years old

“[..] I think it’s so important to realize that and think, Well, if I can’t get the information I need,” whatever it is, “online or through the research going on around me, I can connect with my patient. I can go back to ground zero and build that relationship, ask the right questions, gain their trust, and move forward with that.” --Participant B (Group 1), female, 18-25 years old

Many participants emphasized that, as individuals living with the sickle cell disease, they have expertise in their disorder and the condition of their bodies. They, therefore, suggested the educational advantages of healthcare professionals genuinely listening to their explanations.
“I’ve been dealing with sickle cell for 29 years, so I probably know a little bit more about it than your average doctor. So I think just having doctors and medical professionals just to take time to listen to what the patients are saying and really take it to heart is something really important” --Participant U (Group 5), female, 26-35 years old

Sickle cell disease is highly variable in presentation with variable treatment responses among individuals affected. It is also a condition in which many symptoms cannot be tangibly or objectively measured. Participants, therefore, explained that, as a result, it requires a personalized approach wherein the provider can gain insight from the patient to inform care.

“We as the patients, we live with this disease and we know our bodies better than anybody else. So I know in medicine that there is, like, guidelines people try to follow based on, like, averages. But, like, sometimes that’s, you know, that’s not what works. Because every individual dealing with sickle cell as an individual, we all have in addition to sickle cell disease, we all have other different things that we are going through and things that are going on. And all those factors affect all of us individually.” --Participant P (Group 4), male, 26-35 years old

Many implied that prejudice and stigma may interfere with the providers’ willingness to listen. Assumptions or misjudgments about patients’ needs may effectively inactivate the patient voice, as explained by the following participant:

“We face so much stigma, stigmas between doctors and nurses, because they’re already prejudging us before we’re even triaged. “Oh, she’s here again for pain. It’s nothing wrong with her. Oh, she’s here. She’s in sickle cell crisis. We’re going to make her wait four hours, or five hours.” So we're prejudged before our blood even come back. So my thing would be for them to just listen. Hear us out” --Participant L (Group 3), female, 26-35 years old
A3.1 Walls Interfering with Relationships in Healthcare

The majority of participants conveyed attachment to and trust in their healthcare providers who specialize in sickle cell disease but reported less positive experiences with non-specialist healthcare professionals. Participants in two groups emphasized how experiences of discrimination or cultural differences in healthcare can produce barriers, or “walls,” that interfere with their relationships and interactions with professionals. In Group 2, for example, participants discussed how these walls can occur especially when working with a new provider and worrying about prejudice:

“So, like, when you get that new doctor and you get that new face, you kind of just put, like, this wall up a little bit, like [...] "Oh, I'm not a drug seeker. I'm here for a legit reason.” -- Participant D (Group 2), male, 18-25 years old.

The theme of walls interfering with clinical interactions was also reflected in the discussion with Group 1. Participant C explained how walls are more likely to come up when there are cultural differences between patients and providers:

“If you're working with that population, but you haven’t grown up in those type of neighborhoods, or you don’t understand lingo or just the culture in general, it’s going to be very hard for that clinician to actually break through to even understand. And, so, this barrier and wall automatically comes up” --Participant C (Group 1), female, 26-35 years old

She added that in these situations providers must be willing to be “flexible” and recognize biases to “break down” the walls by asking questions to gain a better understanding of their patients. In the same group, Participant A conveyed the importance of gaining awareness of the group-based challenges experienced by the patients to achieve a better overall understanding of them:
“Find out what issues that they are dealing with outside of your office, so that way you can be more understanding and actually have that to draw on when you actually meet the person. So, researching your demographic before you meet the person” --Participant A (Group 1), male, 26-35 years old

In sum, whether it is due to stigma, cultural barriers, or social differences, participants perceive that walls interfering with clinical interactions and relationships are largely the result of feeling misjudged or poorly understood by clinicians.

A3.2 Racial Concordance Can Facilitate Connection, but the Role of Race is Complex

In groups where the topic arose, participants were asked if and how race, ethnicity, culture, and age of the clinician influences the quality of patient-provider interactions and relationships. Responses were varied and nuanced.

A minority of participants explained that they perceived the social factors of a clinician, such as racial identity, to have an impact on their interactions. Some shared examples to support their opinion as to why the race of the clinician makes a difference in the interaction or relationship. For example, Participant Q stated that, apart from his current hematologist, “black people treat sickle cell better.” He explained that differences in interactions stem from differences in clinicians’ ability to empathize with their patients, providing an example of different experiences in parallel situations. He compared seeking emergency care in a small demographically homogeneous Texas town to receiving emergency care in Nairobi, Kenya. He said he perceived significantly better care and attention at the Nairobi clinic, despite it having significantly fewer resources. Participant Q provided another example of how, in the U.S., he would wait to go to the hospital during the evening shift when the African-American nurse was working:
“I knew the African-American nurse worked the night shift, so if I was sick in the morning, I'd stay home in pain, waited till her shift was on before I went to the hospital. Because when I got there, it was automatic for her, you know. She got it. Meanwhile, you know, the Caucasian nurse, she wants to help but, you know, there's a limit to her understanding of the situation. And I get that. I'm not judging her for that. There is a limit.” --Participant Q (Group 4), male, 36-45 years old

In the same group, Participant O shared that although he had not experienced discrimination by clinicians of other races, he commented that African or African-American providers and staff had made relatively more of an effort to get to know him on a personal level:

“She took a liking to me and I of her and just the staff themselves got to know me on a personal level and I never had that experience with any of my doctors, even here at [HOSPITAL]” --Participant O (Group 4), male, 36-45 years old

In another group, when participants were asked in an open-ended way about how they think genetic counseling could be useful or harmful in the context of sickle cell disease, Participant C responded that she believed in the importance of patients of color being able to identify with their genetic counselors:

“I also would like to say that when it comes to dealing with individuals who are potential carriers-- because we're talking about minorities here, it's important that there are genetic counselors who look like them” --Participant C (Group 1), female, 26-35 years old.

In contrast, when asked if the role of race, ethnicity, culture, and age of the genetic counselor would be important, most of the other participants deemed other attributes of the clinician, such as knowledge of sickle cell disease and empathy, as priorities:
“What matters to me is the practitioner’s willingness to understand sickle cell, to research it, to learn about it to feel comfortable talking about it. So that’s number one with me, knowledge base. And right up there is the person’s capacity to feel, to empathize, to understand, to try to connect with me or, you know, their patients. To say that, you know, even though I don’t have the disease or I don’t have a family member with the disease, I can feel for you, I can understand, you know, how you must feel. Or, you know, just that heart, that’s what I’m looking for, knowledge base and heart” -- Participant R (Group 4), female, 46-55 years old.

Other participants such as the two below discussed that while racial discordance does have an impact, barriers can be mitigated with the right effort. For example, Participant C also discussed the importance of bias training and culturally-sensitive communication in mitigating the effects of racial discordance:

“And then, again, have training. I mean, anyone can build relationships with anyone. We’re humans. It’s just making sure you have that training on how to talk to individuals from different cultures. You know, it’s not that hard” -- Participant C (Group 1), female, 26-35 years old

“I agree. It’s not that I’m only going to connect to someone if they’re an African-American female. Like, I think you can build a relationship with any other human. It takes understanding, it takes empathy. It takes all those things” --Participant B (Group 1), female, 18-25 years old.

Similarly, the participants in Group 2 suggested that race indeed has the potential to lead to barriers. However, openness, communication style and receptiveness of a clinician to listen to the patient can help to diminish the barrier.

“So, like, you know if I’m there for, like, sickle cell counseling and I see a white person walk in the room, immediately, I’m
going to, like, kind of be, like, okay, like, they don’t get it as much kind of thing. Though, you can still overcome that barrier by how they are open, how they communicate with the person, how willing they are to, like, receive information like that. So yes and no. Like yes, it could play a part, but it’s something that’s very easily-- a wall that can be easily torn down” --Participant D (Group 2), male, 18-25 years old.

He also added that racial concordance is neither a safeguard from prejudice nor a guarantee for relatability, providing examples of how some of the most discriminatory providers he has encountered have been African or African-American, and some of his favorite nurse practitioners have been White. In sum, responses indicated that racial concordance/discordance certainly has the ability to influence interactions and relationships in the healthcare setting, but the influence is likely nuanced, complex, and contextually dependent. Common within participants’ responses was the need to feel understood and empathized with by healthcare professionals.

A4.0 “People Don’t Talk about Sickle Cell:” Low Awareness in the U.S.

A4.1 Lack of Knowledge about Sickle Cell

When participants were asked how they believe the general public perceives sickle cell disease, the consensus across all groups was that society doesn’t think or talk about sickle cell in the United States. On a societal level, a poor understanding of SCD translates to a poor understanding of the challenges individuals with the disorder face and/or social stigma. Even among African Americans, participants believed that there is still limited awareness of and attention to the condition.

“Even certain blacks in the same community that you live in, they don’t know. You have to educate them, you know. I mean, one of the key factors is once you say it’s a blood
disease, they think they can get infected. It's not that way; it's hereditary.” --Participant P (Group 4), male, 36-45 years old

“And there are black celebrities who don’t even donate to sickle cell to help their own people” --Participant E (Group 2), male, 46-55 years old.

A few reported that even within families of individuals with sickle cell disease, there remains a limited comprehensive understanding of the condition:

“I thought my dad, being that he had two siblings that have died from it, he knew my struggle. I’m still educating him every single day” --Participant K (Group 3), female, 36-45 years old.

Participants in this study attributed the low awareness of sickle cell disease to its status as a “minority disease.” In addition to a history of receiving low funding, sickle cell disease does not receive the same amount of media or public health attention that other chronic conditions do. Furthermore, participants within all five focus groups expressed that lack of awareness about sickle cell disease among members of society and healthcare professionals is partially attributable to its absence from education.

A4.2 Low Awareness Among the Healthcare Community

Misinformation can also contribute to the stigmatization of sickle cell patients in clinical care. In four groups, participants raised the point that sickle cell disease is not given much attention in the education and training of health professionals who will need to understand sickle cell disease in their practices. This lack of knowledge could translate to misinformed care or stigmatization of patients in the clinical setting.
“And I think, like, the biggest sign of the prejudice that exists in the medical community, or not even necessarily prejudice but it is prejudice, too, but the miseducation about it in, like, the medical profession is-- it is my biggest pet peeve when I’m getting, like, the nurse is about to give me my IV and we’re making conversation and she asks, ‘So how long have you had sickle cell?’” --Participant D (Group 2), male, 18-25 years old

“And, like, I have a friend who is a nurse and she actually said she had to correct one of her nursing students when they went over sickle cell, and because one of the nursing students was like, "Oh, yeah. You know, they come in a lot, you know, for drug seeking." And this was, like, a couple years ago. This is not, like, a long time ago.” --Participant G (Group 2), female, 26-35 years old

A4.3 Early Education Plays a Role in Public Awareness

A few participants mentioned that education about sickle cell disease is also limited in primary and secondary educational settings. In focus group 5, for example, one participant brought up how sickle cell disease is used in primary education to teach students about the basics of genetic inheritance. However, rarely are students taught the implications of sickle cell disease. Individuals in the same discussion agreed that it would be helpful from an awareness standpoint for students to be educated about what sickle cell disease entails for individuals living with the disorder as well as how it is treated:

“First of all, talk a little bit more about how sickle cell can affect individuals-- okay, a lot of times what’s talked about is the genetic inheritance, which is fine, but if you’re going to talk about that, at least add in there, "Okay, and people who inherit this disease can have these particular symptoms or conditions." --Participant U (Group 5), female, 26-35 years old
Another participant in group 5 added that, during his early schooling, sickle cell disease was discussed with levity, which had caused him to believe that his own condition was comparable to asthma or the flu in terms of severity:

“In school, we had learned about cancer in like sixth grade and what it was, and how many different kinds of cancer there is, so I sort of just thought that sickle cell was sort of on the same level as like asthma or people getting the flu. So it confused me, because I didn’t know how severe it was. Like people tell you things as a child, but you don’t really know things. You’re sort of just there kind of doing things but you don’t really know what you’re doing or why you’re doing it. And so, I thought that-- I don’t know, I just-- I didn’t think that sickle cell was a big deal. I think other things like asthma were sort of bigger and things you have to worry about more” --Participant S (Group 5), male, 18-25 years old.

B. Dimensions of Decision-Making

To frame our discussion about genetic counseling, we asked participants about their general experience related to reproductive decision-making and having children of their own. The following themes capture the psychosocial dimensions of individuals’ decision making: (1) Individuals Have to Weigh Multiple Risks and Uncertainties (2) Loss of What Could Have Been (3) “I’m a Mother in a Way”: Individuals Pursue Multiple Avenues of Parenthood.

B1.0 Individuals Have to Weigh Multiple Risks and Uncertainties

B1.1 Pregnancy Risks in Sickle Cell Disease

Across all five focus groups, participants explained the impact of serious pregnancy-associated health risks for women with sickle cell disease and their unborn children. Women in the group who had experienced pregnancy shared accounts of life-
threatening events while pregnant. For some participants, these events were unforeseen by both themselves and by their obstetric providers. Women moreover stated that they were uncertain whether the medications they take for sickle cell would pose serious risks to the pregnancy. Many of the women who have not yet had children stated that health risks associated with pregnancy are a significant contributing factor to their uncertainty about having biological children in the future:

“I would have to get off medication in order to even get pregnant and carry a healthy child. So that’s another step that I have to think about and, ‘Okay, if I do pursue this thing of having a family, then how am I putting myself at risk by having to get off all my medications, and how does that change my life?’” --Participant U (Group 5), female, 26-35 years old

**B1.2 Partner Choice and Risk of Recurrence in Future Generations**

Across all five groups, participants brought up a concern with passing on sickle cell disease to their children. Several of the younger adults expressed worry of recurrence of sickle cell disease in their children, and participants past reproductive age expressed experiencing similar concerns in earlier adulthood. Some expressed that it would be too great of a risk to have a child who would endure the same challenges as they do, with potentially worse severity. One participant articulated the challenges she and her partner currently face with this issue, as her partner was tested to have sickle cell trait:

“I don’t have any children yet. My boyfriend has trait. I have the full SS. And if I’m completely and totally honest and transparent, it scares me to death to have children. Even though I have the disease in the manner in which I have, where I’m able to hold a full-time job, I’m able to go out with
my friends, I'm able to pretty much maintain a fairly normal lifestyle-- I might be a little bit slower at times, but for all intents and purposes, I'm able to maintain a healthy and normal lifestyle. But I can't pick that for my child. I can't-- I'm about to tear up. I can't knowingly give my child something and I don't know how it's going to manifest itself” --Participant J (Group 3), female, 36-45 years old.

Additionally, several participants stated that a future partner's sickle cell trait status would be a non-negotiable deciding factor in their choice of relationships. Others stated that they would not risk having a child who has their disorder:

“But, like, Participant C said, it's tough, because when you’re first getting to know someone, it's like, ‘Do you have the trait? Do you have sickle cell?’ And these are preliminary things that are non-negotiables in any relationship I pursue” --Participant B (Group 1), female, 18-25 years old.

“Do you see how much my life is just chaos? You know what I'm saying? I was born and bred Catholic. It's all I know, and abortion is not an option, but to me, I'm never going to bring a child born with my disease, knowing what I went through, even if there are odds of curing it” --Participant K (Group 3), female, 26-35 years old.

A number of individuals noted that there is a lack of feasibility in ensuring potential romantic partners have non-carrier status, as asking for someone’s carrier status is not considered normal behavior when meeting other people:

“And the thing is, my family, "Oh, you need to have a kid, blah, blah, blah," and I say, "So you want me to go out, and any woman I meet, ask the woman, 'Are you AA? Because I'm SS'" --Participant H (Group 3), male, 56-65 years old.

Although the vast majority of individuals in this current study shared that they made a choice not to take the risk of recurrence in the next generation, reproductive decisions
are personal decisions and vary from individual to individual, regardless of that individual’s disease status. One of the participants suggested that the desire to have a child may outweigh the risks of having a child with a disability. She had empathized with a comedian on the radio who shared that he doesn’t care if he has a child who must ride the “short bus,” as long as he has a child:

“And I mean, it's funny but not funny, but I was listening to, like, a radio station, and I don't know if y'all have heard of this guy, he's a comedian, but he has a child on the spectrum and he said he went through genetic counseling and he was like, "I don't care if my child has to ride the short bus, I want a baby." --Participant F (Group 2), female, 36-45 years old

B1.3. “I Want Children; I am Just Scared to Have Them”: Unpredictability of Future Health

A topic that arose among both men and women across all five focus groups concerns the uncertainty of the trajectory of their health, which would have a direct impact on their ability to be present to care for their children. A minority of the participants articulated that a decision to have children would necessitate having a strong support system if fallback plans become necessary; most participants in their groups expressed agreement on this.

The variability and unpredictability of the course of sickle cell disease bring on psychological, emotional, and social impacts for those affected. Several participants reported that during multiple times throughout their lives, they had been told information by healthcare providers about their life expectancies that turned out to be inconsistent with how long they have lived:

“Mine [life expectancy] is at 60 right now, because I’m 56. So, they go, 'Maybe you get to 60.’ So, when I pass 60, they
will put it at 63." --Participant H (Group 3), male, 56-65 years old

“And every day they tell my mom, "Oh, she’s not going to live until she sees 16. Oh, she’s not going to live until she sees 20” --Participant N (Group 3), female, 36-45 years old.

More men than women emphasized the impact of uncertainty over their health, as well as uncertainty over their financial stability related to their health, on their plans to have children. One man shared that even if his future partner does not have sickle cell trait ("A"), thereby ensuring that sickle cell does not recur in his child, his own health would not be guaranteed:

“Okay, she’s A, so there’s no chance your kid could get it. Yeah. But I had open heart surgery. I may die tomorrow, like. [...] Am I going to have a crisis? You know, that, you know, that ends up with my waist breaking down or something on my hips and I won’t be able to walk again? So, I don’t know how that helps raising a child. So, the funny thing is, I think I want children, I’m just scared to have them” --Participant Q (Group 4), male, 36-45 years old

One participant explained the challenges he experienced as a father with sickle cell disease, especially because the children are young and do not yet understand the physical limitations his disorder places on him:

“So, I have a two-year-old and a six-month-old. The two-year-old is starting to get it, like, the days that I don’t feel well. You can see it in her face, like her face drops when I can’t play with her like I want to and things like that. So, you know, it’s kind of a hard conversation to say, like, “Hey, I want to play with you, too, but I can’t because my back hurts.” A two-year-old doesn’t get that." --Participant A (Group 1), male, 26-35 years old
B2.0 Loss of What Could Have Been

Overall, many participants expressed feeling or having felt grief from the inability, possible inability, or decision to not have biological children. One woman described a devastating premature birth and subsequent infant death that occurred due to a sickle cell crisis she had experienced. Another woman discussed how she may be unable to have a pregnancy due to her bone marrow transplant. A few other women, such as the following participant, had been given conflicting information and vague explanations about whether it would even be possible for them to have biological children.

“...because I know when I was younger, I used to hear that like, "You can't have kids because of your sickle cell. This is really not an option for you," and it used to really discourage me because as a little girl, that's something you think about, having kids, and it sounded sort of like, "No, because of your sickle cell you can't have children" --Participant T (Group 5), female, 18-25 years old.

Lastly, a few of the male participants conveyed familial pressure for them to have children. One participant who immigrated from Africa explained that back in his home country, men continue the family name. Therefore, if a man in the family does not have children, it could potentially end the family name:

“Exactly. Very difficult. "Oh, your name is going to get lost." I mean, because back home-- you ladies are lucky-- but the male--” --Participant H (Group 3), male, 56-65 years old

“Carry the name” --Participant L (Group 3), female, 26-35 years old.
B3.0 “I’m a Mother in a Way, Too”: Individuals Pursue Parenthood through Multiple Avenues

Participants stated that family and parenthood were very important to their lives, even if they do not have biological children. In two groups, three participants shared different experiences of caring for children who were not biologically their own but nevertheless experience the fulfillment of parenthood because of the parental duties and care they provide to others. Examples raised by participants include providing care for nieces and nephews, mentoring children, and financially supporting orphaned children overseas. These participants explained that although they do not have biological children of their own in the traditional manner, they consider the children they provide for as their own children, in a sense:

“So the thing of it is since I can’t have one of my own, in my own family, the orphans, I put them through school. I take care of their school fees. As long as you want to go to school, I’ll pay for you to go. So I take care of those kids, and since I have so many-- I end up having so many kids, even though I haven’t delivered one myself, my sisters and brothers, there are so many of them. So I’m a mother in a way too, and a grandmother. They call me Auntie Grandma” --Participant I (Group 3), female, 66-75 years old.

“I worked with kids before. So that’s where my heart is at. So, I can work with kids again and just mentor them in some type of way. That gives fulfillment, you know. I could add them on social media and see how they’re growing” --Participant O (Group 4), male, 36-45 years old

“That’s why when I leave I have to go and pick up my niece because my sister works in [CITY] [...] so I do this for her Monday through Friday [...] So, I actually have kids” --Participant H (Group 3), male, 56-65 years old.
Furthermore, multiple participants also emphasized that having children biologically was not the only way to become a parent. Some participants noted that while they may not have biological children, they would consider, for example, adopting children. For example, one participant who is undergoing a bone marrow transplant, a curative procedure for sickle cell that can compromise patients' reproductive ability, expressed that she is open to adopting and has a specific interest in a program in which adoptees are children with disabilities:

“But again, I'm very open to adoption, and the reason why I do nonprofit for adoption is because I have a unique story where I'm an immigrant-- you know what I mean? And Gift of Adoption is specifically for couples that want to adopt kids with disabilities” --Participant K (Group 3), female, 26-35 years old

Another participant who has not yet had children shared that having a family was very important to her. She shared that she will have a family of her own through some means, regardless of whether she will be able to have children through pregnancy. She believes it is important for healthcare professionals to share with patients viable options that a patient could consider to have a child, rather than exclusively discussing the risks of pregnancy in sickle cell disease.

“Because it's easy to see fear. It's easy to see, like, what could happen, like the precautions, whatever it is. But, I guess, constantly reminding them, but 'If a child is what you want,' showing them other options, like, “you don’t have to do pregnancy. There are other routes.” I think it’s important to just make sure you're maintaining that positive outlook” --Participant B (Group 1), female, 18-25 years old.
C. Experiences and Perspectives on Genetic Counseling

Participants were asked about their experiences with and perceptions of genetic counseling. Most participants in this study had not received genetic counseling and did not have a comprehensive understanding of what genetic counseling involves. They, therefore, provided their responses based on the experiences of other participants and/or the definition of genetic counseling provided by the facilitator, which can be located in Appendix E.

C1.0 Genetic Counseling is a Way to Gain Knowledge and Learn Options

Most of the participants in this study expressed that they had a good understanding of the genetics of sickle cell disease, including the nuanced details (e.g. there are multiple types of sickle cell carrier status). However, most of these individuals stated that they acquired this information from their own research or through their parents versus in the clinical setting.

"Everything that I was taught was, like, the genetics came straight from my parents and the internet" --Participant P (Group 4), male, 26-35 years old.

Despite having a clear understanding of the genetics of sickle cell disease, participants shared that they do not know what reproductive options are available to them and would benefit from learning about them.

The majority of the participants in the study responded that genetic counseling would not be relevant for them currently. This is because they considered genetic counseling to be useful only in the family planning and reproductive context. Nonetheless, participants generally stated that they believed genetic counseling and
genetic education related to sickle cell disease are important. A typical response was that the information provided could be helpful to future parents to make informed decisions and prepare for their next steps. These participants explained the relative value of having information to move forward with having a family:

“I think it’s very, very important for anybody considering having kids to see a genetic counselor and to find out exactly what diseases their child could present with, once that couple has decided to have a kid. And then, also, bringing us into sickle cell, that-- I feel-- and I could be wrong about this-- but based on what I’ve heard from other people, is that not all genotypes of sickle cell are tested in the newborn screening or pre-screenings for infants and for babies” --Participant A (Group 1), male, 26-35 years old

“I think it’s very useful, because before, parents, if you put them in a position where they didn’t know, there was not enough information there to help them” --Participant I (Group 3), female, 66-75 years old

“I think it’s really important to take advantage of it-- I mean, useful to take advantage of it-- but I never really considered it outside of having a family. It’s not really something I see necessary if you don’t plan on having a family. I could be wrong, but that’s just how I see it” --Participant T (Group 5), female, 18-25 years old.

In Group 3, participant J responded that she believes genetic counseling should be an option available to everyone, despite their genetic disease status. She explained that it allows individuals to have the benefit of making informed decisions.

“I think it's very useful and I should certainly hope that Participant L didn't get it [genetic counseling] just because she has sickle cell, although it's very important in the sickle cell community. But I think that it should be offered to everybody that's planning on having a child because it can
basically set you up to decide, like she said, whether or not you want to continue on with the pregnancy, and so on and so forth. But especially for sickle cell patients, because, like I said, my mate has the trait, so it’s a higher chance of my child, if I choose to have one, having the disease, and it needs to be offered to everybody” --Participant J (Group 3), female, 36-45 years old.

Four participants reported on the demographics and survey questionnaire that they have met with a genetic counselor or a genetic specialist. Three participants shared their experiences during the focus groups. Of the three participants, Participant L from Group 3 and Participant R from Group 4 expressed that they benefited from having received genetic counseling. Participant F from Group 2 shared a more critical perspective, which will be discussed later. Both participants responded that they appreciated the clarity of the information delivered regarding the pregnancy’s risk for sickle cell disease. Both participants also expressed that the information they received during their genetic counseling visit helped alleviate their concerns about whether their future children would be affected. Generally, participants in this study had not mentioned whether they believed genetic counseling would be useful outside the reproductive context. However, participants did express a clear need for healthcare professionals (genetic specialists included) to discuss or at least take into account the social and psychological challenges the population faces when communicating with patients.

C2.0 Genetic Education Can Help Correct Misinformation

In providing their perspectives on genetic counseling, participants discussed that they believed that genetic education could help clarify misconceptions about sickle cell
disease. Several participants shared that outside of the Western world, many communities still believe in myths about how sickle cell disease occurs. One participant shared that in the U.S., people may be unaware of their trait status, but people generally understand that sickle cell disease is inherited.

“I think from a non-Western perspective when you get into the Global South, you’ll probably find, like, a lot more mis-education, representation of how diseases, but particularly how sickle cell is spread, especially like in Nigeria, in the Congo. Like, a lot of those individuals still believe it’s a curse that’s running through your family” --Participant C (Group 1), female, 26-35 years old.

Individuals who immigrated from West Africa shared that their parents’ generation had very little understanding as to how sickle cell disease occurred. They also discussed that lack of knowledge combined with customs of intrafamilial marriages in some communities put couples at high risk of having children affected with sickle cell.

Furthermore, participants across three groups emphasized that although people may have basic understanding that a couple in which both individuals with sickle cell trait have a chance of passing on sickle cell disease in the next generation, it is not common knowledge that individuals with carrier status for other hemoglobinopathies also have a chance of passing on sickle cell disease (depending on their partner’s genotype).

C3.0 Barriers to Genetic Counseling: Areas to Address

C3.1 Genetic Counseling is Irrelevant to the Youth

Across four focus groups, participants noted that there are challenges leading to limited exposure and, therefore, access to genetic counseling. They raised how even
though genetic counseling may be beneficial, it is not a well-known service for patients. Further, it is likely not a service that people believe would be relevant to them. Most participants shared that they had not ever heard of genetic counseling or had only known about it very minimally. For example, one participant noted that genetic counseling would not be a priority of individuals if they are healthy and have no apparent cause to think about genetic counseling:

“Because when you’re young, and you’re first starting a family, you know, you think you’re young and you’re invincible and that you can you can’t really pass on anything if you live a relatively normal life. It’s when you start getting older, then you start hearing more about genetic counseling.” --Participant C (Group 1), female, age 26-35

Participant C added that healthcare professionals should increase awareness of the availability of the service and its potential relevance to people. She suggested that these awareness efforts should provide answers to common questions people may have related to access to genetic counseling:

I think some of the work needs to be done just informing the younger generation of “What is a genetic counselor? How can they be of help? Is it covered under my insurance? Is this something that I have to pay out-of-pocket?” --Participant C (Group 1), female, age 26-35

Some participants expressed ideas similar to Participant U who said that she had heard of genetic counseling, but would not know how to find a genetic counselor. The other participants in her group agreed, adding that they would not readily know how to access a genetic counselor or know if genetic counseling is even accessible for them:

“I’ve heard of genetic counseling before but I don’t know where I would find one” --Participant U (Group 5), female, 26-35 years old
“Yeah. I mean, my mom has discussed a genetic counselor, but I’m not quite sure what exactly they do, and like Participant U said, where to find one, and is it something insurance would cover, things like that. Not quite sure” --Participant T (Group 5), female, 18-25 years old

“I feel the same way. I’ve heard about them. I don’t know really what they are, what they do, or where to find them” --Participant S (Group 5), male, 18-25 years old

Participants believed that genetic counseling is disconnected from the community. A participant in Group 1 emphasized that healthcare professionals should be proactive in making recommendations and making it known to patients that such services exist and are available:

“You know, like, whether you’re a male or female, if you’re talking to your primary physician or anything, you need to be like, “You know what? I’m thinking about starting a family.” The first thing that should come out of their mouth, “Okay, you need to see a genetic counselor. You need to find out if you’re going to take this step, what could happen?” --Participant A (Group 1), male, 26-35 years old

In Group 2, Participant B suggested that genetic counseling could be incorporated in youth-to-adult transitional care programs for individuals with sickle cell disease, but acknowledged that discussion of genetic counseling may require a minimum level of maturity.

Participant G suggested that earlier exposure to genetic counseling could be made possible if it were integrated into already existing programs and classes for the youth. She commented on how with the current interest among the general public in direct-to-
consumer genetic testing (e.g. ancestry testing), for example, people may be interested in also discussing clinical genetics:

“I think all of that, like, family planning and genetic counseling, I think it would be a great topic to teach in, like, health class in high school. And I think information is just good. So, I mean, I think genetic counseling, it just sounds like information that you’re getting and so I think information is always good. Just like with these whole DNA things that are coming out, like find out where your family is from” --Participant G (Group 2), female, 26-35 years old.

C3.2 Resource Barriers in African-American Communities

Participants across four focus groups expressed that services related to family planning are lacking, in general. A participant from another group attributed disparities of resources to the Black community not being adequately informed about health-related resources, in general.

“[...] it is a predominantly black disease and there aren't resources always readily available in the black community, that is also why they could be lacking. Like I said, I’ve never heard of any, but also I haven't been in a family planning position where-- but I think that could be a reason as to why some of us don't know about resources” --Participant J (Group 3), female, 36-45 years old

Participant J contextualized this statement, explaining that certain areas of health do not receive attention in African American communities. This is influenced by historical underpinnings in America:

“I think that there’s a lot of different classes that need to be taught in the black community about even mental health, because it’s a large stigma in our community, where you just don’t talk about certain things. Like black men are supposed to be men and they can't cry and they can't express their
feelings, and I think that's-- the things that we were initially taught, which date back to slavery." --Participant J (Group 3), female, 36-45 years old

In response, Participant I from the same group stated that she believed resources are available as long as one looks for them: “I always think that resources are available if you know where to find it, or if somebody lets you know where it is.” Participant J responded that she agrees but contends that communities are not informed about them: “Again, that's my point. We don't have people telling us.”

In another group, one participant commented on the importance of people having more information about genetic counseling. Similar to Participant C above from focus group 1, she also noted issues with exposure and awareness. She shared her personal experience and questioned whether individuals would have exposure to genetic counseling early enough if they do not carry out family planning in the “traditional” sense.

“As far as the services, so you said the genetic counseling-- okay, so as far as family planning, I didn't do any family planning. We probably don't really do that too much in the black community” --Participant G (Group 2), female, 26-35 years old

“Mmm” <laughs> --Participant D (Group 2), male, 18-25 years old

“Mm-mmm” --Participant F (Group 2), female, 36-45 years old.

“I can't speak for everybody, but” --Participant G (Group 2), female, 26-35 years old.
"No, facts <laughs>" --Participant D (Group 2), male, 18-25 years old

"<laughs> So as far as family planning, so yes, the service was available as far as, you know, getting her father tested. So we, you know, were like, "You know, we’ll maybe have children at some point." But the pregnancy wasn’t planned which, you know, if you talk to my mother, she’s like, "How can that not be planned?" --Participant G (Group 2), female, 26-35 years old

Perspectives on lack of exposure, lack of knowledge about, or lack of utility for genetic counseling reflect possible barriers between genetic counseling and people who may find it useful.

C4.0 Information May be Perceived as Harmful: Manner of Delivery Matters

Whether or not they had had genetic counseling, the majority of participants responded that they could not think of any potential harm directly from genetic counseling. However, one of the three participants (Participants L, R, and F) who received genetic counseling shared a less than positive overall experience from genetic counseling. Another participant who received genetic counseling shared that she had a generally positive experience. However, she recalled one negative aspect of being dissuaded against pregnancy. The interpretation from these responses was that risk information delivered during genetic counseling may feel simultaneously impersonal and threatening to the patient. The manner in which the information is delivered is therefore very important to reducing perceived harm during the discussion.
In contrast to the participants who deemed their genetic counseling experiences as overall valuable, Participant F from Group 2 shared a more critical perspective of her own experience with the service. She discussed that she perceived the information provided to her during genetic counseling was more for “the general masses,” rather than tailored to her specific situation:

“Well for me when I got pregnant, I mean, I knew about genetic counseling from, like, the prior pregnancy and so I decided to go again. But in my experiences, I mean, with it, it wasn’t so positive because I felt like it was really for, like, the general masses and not specifically for my situation. It’s like, "Well, you know, we test for this." But this affects this population and this doesn’t apply to me, you know.” --Participant F (Group 2), female, 36-45 years old

Participant F also expressed that there was emotional harm associated with receiving the risk statistics about her pregnancy in the manner that she did:

“I mean, just to really communicate that nothing is 100 percent. Because like to me, like, to say there’s a 25 percent chance of something or a 50 percent chance of something, if it never happens, then to me, that’s a 0 percent chance, you know. So it’s just like I felt like it’s Russian roulette with people’s emotions in a way.” --Participant F (Group 2), female, 36-45 years old

In response to Participant F’s perspective on her genetic counseling experience, Participant D from the same group agreed that such a discussion can be very personal to a patient. He emphasized that he had a very trusting relationship with his hematology nurse practitioner, noting that he would be comfortable discussing sensitive topics potentially raised in genetic counseling with a provider like her. Participant D therefore, suggested that it may be helpful for the referring provider whom the patient already has
an established relationship with to provide context to the genetic counseling referral beforehand:

“So it would be helpful rather than just throwing another person in that mix and being like, "Oh, okay, and this is someone that’s going to talk to you about genetic counseling," you know, and have someone that’s a part of your, like, core team that you work with regularly to have that conversation with you. And then if they would want it would be like, "Hey, okay. Like, here’s somebody who’s more well versed in it," and then like kind of like refer you to them. But I think it would be really helpful to have the primary person that you’re working with, either your primary care or your hematologist, whichever one you’re seeing more often, to be the one to, like, at least have that initial conversation with you” --Participant D (Group 2), male, 18-25 years old

Participant D added that sickle cell disease is attached to a significant stigma that influences how healthcare professionals perceive patients. He explained that when they meet a healthcare professional for the first time, patients feel pressure to disprove this stereotype. And as a result of the stigma, he explained patients with sickle cell may feel pressure to close themselves off from the healthcare professional.

Participant F responded in agreement, acknowledging that she did not perceive noticeable attempt by the genetic counselor in this case to establish rapport with her prior to a genetic counseling discussion that felt invasive and personal:

“It’s very informational, but I think for me, the walls did go up because it felt invasive. It didn’t feel rapport setting just yet. Because you’re talking about information that’s very personal, you know. So it’s like once that has been established, I believe you’d be more receptive to the information. And then you’ll be able to kind of utilize it. But
once those walls go up and then you already feel attacked” -

-Participant F (Group 2), female, 36-45 years old.

In contrast, however, Participant G from the same group noted that, for her, it would just be important to receive the information and the person delivering the information would be less important.

In another group, Participant R explained that it is important for clinicians to allow the patient to make her own reproductive decisions, even if there is a risk for sickle cell disease:

“Remain neutral. Do not try to dissuade a person with sickle cell from having children. Even in the event that the person, the patient’s spouse or partner has the disease, leave it up to them. I mean, I have sickle cell and I’m okay and I’m happy that I was given that opportunity to live” --Participant R (Group 4), female, 46-55 years old.

D. Views on Racialization of Sickle Cell Disease and on Ancestral Framing

The focus group participants were asked for their views on racialized views of sickle cell disease held by the healthcare community and beyond. The latter part of the final objective of the study was to explore reactions to an alternative ancestral framing to describe sickle cell disease that emphasizes its evolutionary significance throughout history. To achieve this objective, each focus group was asked for their perspectives on the following ancestral frame of sickle cell disease:

“Scientists have discovered that sickle cell trait happened in humans over 7,000 years ago. Sickle cell trait and sickle cell disease still exist today because people with trait have stronger protection from the deadly infection malaria. While the trait gene started in Africa, both the trait and disease can be found in many parts of the world among many ethnicities. This is because of people migrating throughout history. Scientists say the sickle cell trait gene protected the world from malaria.”
The participants were asked for their thoughts on this information and asked if learning the above information changed anything for them. Additionally, they were asked whether they believed there was value in sharing this information with others. Responses were varied, with some participants considering this information significant, others considering it potentially significant in certain contexts, and others considering it to have little significance or impact.

D1.0 Pigeon-holing of Sickle Cell Disease Can Have Ramifications

Within every focus group, participants emphasized that sickle cell disease is unwarrantedly “pigeon-holed” or “stigmatized” as a black disease in the U.S. healthcare setting and in society despite it being well-documented that members of several racial and ethnic populations and individuals of mixed ethnicities are affected. Participant I from Group 3 explained that the world is getting smaller, meaning that modern travel facilitates travel, immigration, and therefore interracial marriages contribute to the applicability of sickle cell disease across racial groups:

“You see, we get stigmatized for no reason, it’s a black disease, but it’s not a black disease. Right now this disease is universal. A long time ago, you sit in a sailing ship, it takes you three months from one side of the Atlantic to the other. Now, within eight hours, you can go from California to Tokyo. So the world is getting smaller, and the diseases are getting smaller. So everything is getting close. --Participant I (Group 3), female, 66-75 years old

Across all the focus groups, participants gave examples of non-African-American and non-Black groups in which individuals are affected with sickle cell disease.
“And I hate the fact that it’s pigeonholed like it’s a black disease when they have even proven and studies taken that the Jewish community can have it. Southern Italians can have it. As far as—” --Participant O (Group 4), male, 36-45 years old

“Indians.”--Participant R (Group 4), female, 46-55 years old

“Indians from actual India, you know, not Native Americans” --Participant O (Group 4), male, 36-45 years old

“And even Asians, thalassemia.”--Participant R (Group 4), female, 46-55 years old

Participants shared that this notion is essentially misinformation that would have negative consequences for African American/non-Black individuals who are affected. One participant shared how drug-related stigma afflicts individuals with sickle cell disease, regardless of race:

“I think it’s negative in the sense that, because, in the United States at least, sickle cell disease is considered an African-American disease, people who aren’t African-American and present are stigmatized as well, because they get to the hospital and they’re in crisis and in pain and the uneducated doctor is going to say, “Well, you’re Caucasian.” Like, “You can’t have sickle cell.” And then they’re put off to the same—like, they’re a liar or they’re a drug seeker, and they’re not. [...] And even as migration happens and we’re moving, it’s going to be more and more people who aren’t considered People of Color are going to be born, especially with interracial marriages, with sickle cell. So, those individuals are stigmatized as well. Just because of lack of information and education. That’s bad, too, because anyone who goes through this understands the pain and I don’t care if you’re black, white, yellow, whatever; when you’re in that amount
of pain, you need to be seen immediately and taken seriously" --Participant C (Group 1), female, 26-35 years old.

A few participants responded that they felt that the stereotyping of sickle cell disease as a “black disease” likely has negative implications but does not personally affect them, noting that in reality, most people in the U.S. who are affected by sickle cell disease are African American or Black. For example, the two participants below from different groups expressed the potential social negative consequences but noted these views do not have a substantial effect on them. They shared that with so many other challenges associated with having sickle cell disease, racialized views of their disease are not a primary concern.

“I think it doesn’t-- It’s bothersome, yeah. I agree with everything everyone said about, you know, because it’s thought of as a black disease the treatment is different. I think I’m a little desensitized to it a little bit only because, like he was saying, there are so many other struggles. And so it just kind of gets lumped into. Because I think it would be different if maybe I lived in, like, a place where there’s less black people. And so if I was, like, in a predominantly maybe white country or place and it was thought of as, like, a black disease, it might be more-- it might affect me more. But because I’m already, like, black, like, everything I do is black. And so the disease is-- it’s just lumped into that” --Participant C (Group 1), female, 26-35 years old.

“Yeah, people think that sickle cell is a black disease. I don’t have a problem with people thinking that because I think that the vast majority of people who have sickle cell are probably black, and it has-- I mean, I don’t know. I guess it can be seen negatively because might think, "Oh, black people are diseased," or whatever, but me, it doesn’t make a difference to whether people think it’s a black disease or not” --Participant U (Group 5), female, 26-35 years old.
Although there have been recent advances in the landscape of treatment for sickle cell disease, sickle cell disease has gone for over a century without targeted interventions or therapies. Other participants expressed that a major ramification of racialized views of sickle cell disease is that it has stifled attention to and research on the condition.

“[...] if most patients with sickle cell are African-American, who is researching the disease? Who is funding the disease? If it only affects African-Americans and people who are funding things are not African-American, it’s out of sight, out of mind.” --Participant B (Group 1), female, 18-25 years old

“I hate to say it, but I genuinely do think part of the reason why even though it’s, like, the oldest genetic disease and we’ve only come so far is because it’s stigmatized as, like, a black disease, and that’s part of the reason why it hasn’t received so much attention and things like that.” --Participant D (Group 2), male, 18-25 years old

“You know, I think that anything that affects us is really--and I say us, black people, anything that affects us I don’t think that it’s going to get as much attention as other ailments or diseases or disorders. I think that other things are so widespread that there’s so many people who buy in and, like, "Oh, it’s, you know, da-da-da-da." With sickle cell, the common knowledge is it affects black people, so there are just fewer people, you know, buying in and trying to help out” --Participant E (Group 2), male, 46-55 years old.

Participants in Group 2 indicated how lack of historical progress is observed in the unchanging treatment of sickle cell-related pain: “Like narcotics, fluids, go home, and then medication is, like, other medications here and there.” They allude to the differences in how the opioid crisis is being viewed and addressed relative to the crack epidemic in
the ’80s and ’90’s, insinuating that the former is not criminalized because it affects White individuals:

“It is like you did not care for the crack epidemic in the eighties [...] It’s just like but now that the opioids are affecting Caucasians, then all of a sudden it’s like it’s not criminalized. You know. It’s just, like, now we need to seek treatment. We have to do a push forward, you know, and it’s just not right. And it’s just like we need to care about the world and everyone” --Participant F (Group 2), female, 36-45 years old

Similarly, participants also expressed the effects that racialized views of sickle cell disease have on affected patients. Participant A from Group 1 alluded to the deleterious consequences of how the media has portrayed the involvement of African Americans in the crack and opioid epidemic and how that relates to societal views of individuals seeking SCD care:

“And there simply isn’t as much care for People of Color as there is for illnesses that affect, generally, white populations. And then I also think that because of the way the crack epidemic and the way heroin and things like that were portrayed in the media, people automatically assume that if a sickle cell patient is presenting-- coming with pain, presenting with pain all the time, that it gets to a point where they’re just drug seeking.” --Participant A (Group 1), male, 26-35 years old

In response to Participant U’s response that she does not perceive a personal impact of racialized views of sickle cell disease, Participant T from the same group expressed that she believes the effect of racialized views of sickle cell patients may not be apparent but plays a role in every aspect related to seeking pain treatment:
“I don’t think we realize it affects us, but because it is characterized as a black disease, I think that plays a role-- a huge role-- in how we’re treated when we go to hospitals, how we’re treated when we need to pick up prescriptions and medicine, with the whole opioid epidemic-- I think that plays a big role in how we get our medicine, how much medicine is given, and things like that.” --Participant T (Group 5), female, 18-25 years old

D2.0 Ancestral Frame has Potential Significance

Some participants expressed that the information presented in the ancestral frame may be valuable in some contexts but did not consider it to be universally valuable. For example, in Group 1, participants discussed that the information may not resonate with members of the general public (e.g. “a basic American”) in a region in which malaria is not prevalent.

“Again, in malaria-prone areas, like in South Asia, South America, Africa, that’s going to make perfect sense, because people are still dying of malaria and they can’t access medicine right now for malaria in the developing world. Now, I don’t know how important that is in a Western nation where malaria is not as prominent” --Participant C (Group 1), female, 26-35 years old.

The group suggested that it might instead be useful for healthcare professionals and researchers to have this information and “open some doors for conversation.” The participants responded that understanding the historical relevance of sickle cell could perhaps help pique research efforts.

“I think from an educational standpoint at the medical school level and the graduate school level that would definitely help clinicians understand where the disease comes from and the history of the disease. It might even spark interest instead of just saying, “Oh, sickle cell disease is a black disease.” No, sickle cell disease is a disease that came about because of
evolution of humanity trying to protect itself from malaria” --Participant C (Group 1), female, 26-35 years old.

“It can make it interesting for people to want to research sickle cell more. But as far, like, in this country, like, it can maybe-- like, it could be a brief explanation, but it doesn’t have much weight as far as going forward. It’s like, ‘Interesting tidbit, this is why sickle cell developed,’” and then continue”--Participant A (Group 1), male, 26-35 years old.

Participant B suggested that learning this information could perhaps remove some of the race-related biases about sickle cell among the healthcare community:

“So, it is an interesting point. I think it’s important for clinicians and those professionals working around the disease to know that, that may remove some bias as to it being a black disease” --Participant B (Group 1), female, 18-25 years old.

Participants from Group 4 responded that the presented information is not personally relevant for themselves. However, they suggested that it may help with the self-esteem of children who have sickle cell disease:

“Let the kids know that, you know. You know, you’re not-- Because of you, because you can't go out to play, he can go out to play, so you're, you're not broken, you're special” --Participant Q (Group 4), male, 36-45 years old

“That would actually be really helpful I think for the kids” --Participant P (Group 4), male, 26-35 years old

Other individuals believed that the information presented was very valuable and suggested that it showed that sickle cell disease exists for a reason. Individuals from only one focus group perceived significant personal value in learning the information
presented with the ancestral frame. In Group 2, one participant expressed that he believed the information was interesting and it was valuable to learn how sickle cell “helped humanity carry on.” He adds that it could be empowering to share with the community, explaining that it provides historical significance for the existence of sickle cell disease.

“I didn't know that and that's actually kind of cool because, like, as much as I hate it, it's like, all right, cool. Like, without this disease, though, like, we humans might, like, you know, I'm going to assume worst case but best case scenario for us is like, hey, like, humans might not have made it without us, like, you know, kind of thing [...] It's like, all right, cool. I have this giant burden to bear. But because of our ancestors that had this thing, like, it helped humanity carry on. So, like, I think that's like actually a really interesting piece of information especially to share within the community because, like, you know, that can be somewhat empowering to, like, some people. Like, you know, just like knowing, like, the historical benefits and being like, "All right, cool. Like this disease sucks but it's here for a reason and it really helped keep-- progressed humanity along, like, you know, like, because who's to say where it would have been without that mutation" --Participant D (Group 2), male, 18-25 years old.

In the same group, another participant agreed, noting that the information has the potential to be empowering, especially for children who have sickle cell.

Participants furthermore suggested the importance for people in the general public to learn this information about sickle cell disease and agreed that it could be a way to educate people on the relevance of sickle cell disease to those who are not affected by it.
D3.0 Relevance of Slavery and Colonialism with Ancestral Frame

In three focus groups, participants addressed the significance of slavery or colonization with the information presented with an ancestral frame. These responses indicate the importance of considering events of oppression in the discussion of the historical events involving African-American and African individuals. In Group 1, one participant expressed that it should be noted that the spread of sickle cell needs to be explained by not just migration, but also forced migration, referring to the history of slavery:

“I think that anywhere the disease is explained and this comes up, I think it should be noted that it’s not just migration, but forced migration” --Participant B (Group 1), female, 18-25 years old.

Furthermore, in Group 2, Participant G also brought up the importance of acknowledging the history of slavery. She says that understanding the tremendous tragedies of their ancestors who may have lived with sickle cell disease as slaves. She concluded that having this perspective can help individuals living with sickle cell today appreciate that they are in a better circumstance and that care exists, even though, in her words, it “still needs work”:

“I think just knowing, like, which each generation just having a thought of what the prior generation went through is good. Because I mean, I think about, you know, how bad it is for us. But I think about like back in like I think about, like, the slave trade. And when they were on those boats for--” --Participant G (Group 2), female, 26-35 years old

Participants in the same group expressed agreement with the unfathomability of suffering a sickle cell crisis while in capture.
“I could not imagine and just not being-- having any help at all and having to work through that. Like having to work in the hot sun in a crisis. And they don’t care. Like, what? [...] But, yeah, so that makes me, like, appreciate, you know, even the care that we have, even though it still needs work, but it makes me, like, appreciate my situation more. So, yeah, I think history definitely is helpful. --Participant G (Group 2), female, 26-35 years old

Lastly, in Group 3, in response to the ancestral frame presented, Participant I and Participant H brought up the historical significance of malaria driving out the British colonizers who took over West Africa in the 19th century:

“But at the moment, like the scientist indicated, if you have a trait, you are protected, but then you see there’s a joke in Ghana, West Africa, when the British were there and they never left, but then eventually after they leave, people say, "Oh, the malaria drove them out. We couldn’t drive them out but the malaria did <laughs>" --Participant I (Group 3), female, 66-75 years old

D5.0 Ancestral Frame is Not Relevant: Malaria is Still Prevalent and is Harmful to those with Sickle Cell Disease

The majority of participants in Groups 3 and 5 stated that they did not believe that the information about the link to ancestry would be impactful to learn. They noted that this is especially true for individuals with sickle cell disease because malarial resistance does not apply to individuals who have sickle cell disease. For example, in Group 5, Participant H shared his discussion with his physician about how individuals with sickle cell disease are not resistant to malaria without the help of the preventative drug
chloroquine that every other individual would require. In fact, individuals who have sickle cell disease are more susceptible to adverse outcomes if they contracted malaria.

“So I ask him, "How are we getting the protection about this malaria thing?" And he said-- so he ask me. "So when you were in Ghana, did you get malaria?" I said, "No, but my uncle was a doctor and told my dad to give me chloroquine." So I was taking chloroquine every Saturday, and that is what [PHYSICIAN NAME] said, "That is what saved your life." So it's not like automatic there, where you have the sickle cell, you are-- no" --Participant H (Group 3), male, 56-65 years old.

“I was just about to say that" --Participant K (Group 3), female, 26-35 years old.

“Yes. So the chloroquine, it act like an inhibitor for you. If not, you get malaria and you will die" --Participant H (Group 3), male, 56-65 years old.

Likewise, Participant U from Group 5 noted that malaria still afflicts many people around the world today and that she believes it is rather an “overstatement” to say that sickle cell trait has a global protective effect:

“No, I don't think that that piece of information is relevant because, okay, yes people with sickle cell trait may be more resistant to malaria than those who don't have it, but people with sickle cell disease can still get malaria, so it's like, "Okay, fine." Second of all, a lot of people still get malaria every day and people die of malaria every day” --Participant U (Group 5), female, 26-35 years old
DISCUSSION

The study sought to gain a better understanding to enhance the benefits and mitigate the harms of genetic counseling in the sickle cell context by exploring the experiences of individuals with sickle cell disease. A qualitative approach was undertaken with 21 participants divided into five focus groups. Discussions were audio-recorded and transcribed and analyzed through thematic analysis for recurring topics, ideas, and patterns of meaning. We gained deeper insights into how the common healthcare experiences and perspectives of individuals with sickle cell disease influence their preferences in interactions with new providers, such as genetic counseling professionals. Findings suggest that individuals seek collaboration with healthcare professionals and that collaboration is facilitated by effective communication. Barriers may interfere with healthcare interactions and relationships as a result of patients’ previous experiences with stigma, cultural differences, or perceived unwillingness of healthcare professionals to listen due to negative attitudes towards patients with SCD. Findings also demonstrate that individuals with sickle cell disease may have both educational and psychosocial needs related to reproductive decision making. We learned that individuals perceived value or potential value in genetic counseling, however, expressed concerns and questions about exposure and accessibility, particularly for the youth and the African-American community. Lastly, we captured how individuals perceived mostly negative consequences of racialized views of sickle cell disease in the healthcare community and beyond. Our alternative ancestral framing of sickle cell disease had received mixed responses on its perceived value for the sickle cell
community, but participants suggest that presenting the information may be useful in alternative contexts.

Views on Awareness and Perceptions of Sickle Cell Disease

In both the healthcare and community setting, participants attribute negative attitudes towards individuals with sickle cell disease to an interplay of ignorance about the disease and a gap in empathy for individuals affected. Recounting their experiences in healthcare, community, and even familial contexts, participants consider there to be low general awareness/understanding of what is entailed for individuals affected with sickle cell disease. Poor understanding of sickle cell disease within individuals’ communities or the general public can lead to social stigma (Royal et al, 2011, Sankar et al., 2006, and Ola et al., 2016). In clinical settings, it may contribute to healthcare professionals’ negative attitudes towards patients, which can affect the provision of care and patient outcomes. Furthermore, although the historical lack of treatment options for sickle cell disease is most likely due to reasons related to profitability, low awareness of sickle cell disease can lead to its neglect in government funding decisions and research efforts (Bahr and Song, 2019).

From the perspective of participants, attitudes towards individuals with sickle cell disease are in part related to the ability of others to empathize with those affected. Empathy is a multifaceted and complex concept, described in the literature in multiple ways and conceptualized differently depending on discipline (e.g. medicine, nursing, psychology); empathy means different things to different individuals (Jeffrey, 2016). In this study, participants seemed to equate empathy to the ability to understand the
challenges of others and respond or behave accordingly (verbally and/or through action). Racial bias was considered an obvious key factor, as individuals noted examples of neglect of pain or suffering of African-American or Black individuals in general. Furthermore, their accounts of long emergency department (ED) wait times, especially compared to White patients, are consistent with large-scale studies on ED wait times of patients with SCD (Haywood et al., 2013, Pulte et al., 2016). Nevertheless, participants insinuated that negative attitudes or lack of compassion towards those with sickle cell disease are not explained by racial biases alone. This is supported by the observation that stigma of sickle cell disease still occurs in countries in which populations most affected belong to the racial majority group, such as countries in West Africa. Even in the U.S., studies have shown that race alone does not fully explain disparities in care experienced by individuals with sickle cell disease (Haywood et al., 2013; Haywood et al., 2014). Participants in this study suggest that the lack of relatability and/or the invisibility of the disease both interfere with people’s ability to understand its consequences and empathize with those affected.

Moreover, some individuals suggested that lack of awareness or knowledge among healthcare professionals may be due to lack of emphasis on sickle cell disease in their education. They note that gaps in knowledge about sickle cell disease leads to a poor understanding of the people affected by it and what the disease involves. Findings from an intervention study by Beach and colleagues (2015) have demonstrated that among a regional sample of pediatric healthcare providers, educational and experiential videos on the patient experience with sickle cell disease significantly decrease negative attitudes and significantly increased positive attitudes based on pre/post-test measurements.
(Beach et al., 2015). Studies by Haywood and colleagues (2011) and Jenerette and colleagues (2016) have demonstrated similar results with educational interventions (Haywood et al., 2011, Jenerette et al., 2016). These findings are consistent with the perspectives of participants of this study on how ignorance about the disease leads to misconceptions which ultimately lead to negative attitudes towards patients. This highlights a need for future research to elucidate the outcomes of intervening earlier, perhaps while health professionals are still in training.

From a public awareness standpoint, several of the young adult participants (age range 18-25) held that sickle cell disease could be covered in more depth in early education. A few shared that compared to other chronic illnesses taught, sickle cell disease is discussed only very briefly. Because sickle cell disease is commonly used as an example to teach students about genetic inheritance, curriculums have the opportunity to teach about sickle cell disease more comprehensively, covering the challenges people with sickle cell disease experience and ways in which it is treated. Day and colleagues (2015) implemented an experiment using a 5-lesson instructional unit among racially, academically, and socioeconomically diverse students (N = 87) in Oakland, California. The unit included education on genetics and inheritance as well as sickle cell disease. It included information to help break misconceptions about sickle cell disease and provided introductions to careers related to biomedical science. Findings showed that the unit was well-rated by teachers and significantly improved students’ scores on a test that covered science concepts, the scientific process, lifestyle choices, and careers. With recent burgeoning advancements towards sickle cell treatments and therapies, now may be an apt time to bring sickle cell disease into the public conversation in science.
education. Sickle cell specialists and genetics professionals could collaborate with schools to help tailor programs that advance genetics education while also raising awareness on sickle cell disease and increasing exposure to biomedical or healthcare careers.

Lastly, we explored the perceived impact of racialized views of sickle cell disease. The groups agreed that among both the healthcare and greater community, sickle cell disease is seen as “black disease,” despite common knowledge that it affects many different populations globally. They believe that the pigeon-holing of sickle cell disease as a “black disease” decreases the societal attention it receives. Other diseases, such as HIV and cancer, receive greater attention and funding because they are considered to be widespread across many different populations. In the healthcare setting, participants express that racialized views of sickle cell disease have obvious ramifications. There is limited research on whether these perceptions are associated with negative attitudes of sickle cell disease and affected individuals, and findings have been complex. For instance, in one experiment with diverse respondents from the general public (N=1,250), Bediako and King-Meadows (2016) found that respondents who held racialized views about sickle cell disease (e.g. sickle cell disease is a “black disease”) were more likely to have positive perceptions of the disease, but at the same time were less likely to report that they support government funding for sickle cell disease causes.

We received mixed reactions to an ancestral framing of sickle cell disease that emphasizes the protective role of the sickle cell variant throughout evolution. This information was intended to convey the fact that the sickle cell mutation occurred seven millennia ago, when there were around only 5 million humans living on the planet,
and has persisted until the present day. The majority said that the information did not resonate with them personally. A few participants believed this ancestral framing of sickle cell disease was empowering and could be important for members of the sickle cell community to learn, but also for the general public to know. Some indicated that it had potential significance in other contexts and/or for different audiences. Specifically, participants suggested that the information may be useful to help with self-confidence among children with sickle cell disease. Some suggested it may be useful in areas of the world where malaria is pervasive or among healthcare professionals or academics in-training to learn to mitigate racial biases about sickle cell disease. We observed that a group’s responses to this question seemed to be homogenous within the group, suggesting the possibility of groupthink effect. In other words, perspectives on this question seemed to mirror the perspective of the first person who responded to the question. It was also noted that participants may have not understood the point the moderator attempted to make with the information presented, which may be a result of ineffective question wording. Future explorations of ancestral framing should include participants with SCD in the development of the language used to provide the frame.

Overall, participants generally held neutral to very negative views regarding racialized views of sickle cell disease. However, further research is necessary to determine the appropriateness of employing the evolutionary information presented in this study as an ancestral frame.
Healthcare Interactions and Relationships

Individuals in this study expressed that it is important for healthcare providers to acquire better understanding of sickle cell disease; however they also conveyed that the disorder is difficult and complex to treat and that solutions may not be readily available. Knowledge and effective communication cannot replace structural changes needed to take place to address barriers to quality care of individuals with sickle cell disease. However, participants stressed that an understanding of sickle cell disease and effective communication are key to collaboration between patients and professionals.

Individuals in this study also conveyed how barriers in communication can arise between patients and healthcare professionals (HCPs), particularly with HCPs with whom patients do not have established relationships. Emphasis was placed on healthcare professionals’ unwillingness to listen or trust the veracity of the patient experience, which is consistent with large-scale, national assessments of disparities in quality healthcare provider communication (Haywood et al., 2014). Participants expressed the value of healthcare providers establishing a relationship and listening to patients in an effort to work together towards treatment and management solutions.

Individuals also conveyed how barriers in healthcare interactions stem from reasons beyond healthcare providers’ unwillingness to listen. Participants in one group explained that patients may put up emotional “walls” as a self-protective measure due to past discriminatory or stigmatizing experiences in the healthcare context. Other participants stated that racial, social, and cultural differences between patients and HCPs could also contribute to barriers to interactions and relationships. They suggested that in these situations, effective communication with patients and demonstrating genuine
efforts to establish relationships with them ("asking the right questions") are essential to mitigating barriers. Participants in another group suggested the importance of bias and cultural training for clinicians. An overarching theme underlying participants’ responses was the need to feel understood and build partnerships with HCPs.

**Educational and Psychosocial Dimensions of Reproductive Decision-Making**

We explored participants’ experiences regarding having children of their own. Responses across all five focus groups suggested that participants in this study had a correct basic understanding of sickle cell genetics and information about the chances of recurrence in the future generation. Some participants explained that their knowledge of genetics resulted from self-research, advanced education, parental guidance, or pediatric-adult transitional programs.

Reproductive decisions are deeply personal to the individual and education plays only one role among many factors. Our participants communicated several overlapping challenges they have experienced or are experiencing when asked about issues with deciding to have children. Both women and men had expressed concerns over their own health, fears about a recurrence of sickle cell disease in their children, and pressure to ensure that they find a partner who is a non-carrier. Women shared personal experiences of life-threatening scenarios of pregnancy. These findings mirror the challenges reflected in prior studies, such as those by Gallo and colleagues (2010) and Rance and Skirton (2019). Women also expressed receiving conflicting information about whether they would be able to have children because of their sickle cell disease, which both
complicates their planning and can have detrimental psychological and emotional effects on girls and women.

Because the vast majority of prior studies on reproductive decision making in the sickle cell context have focused only on women, our findings shed light on male perspectives on this topic. Several men across focus groups experienced pressure from family and friends to have children of their own while struggling with uncertainty over whether they would be able to physically and financially to provide care for their children.

Some participants implied that they have adapted to their own possibility of not having biological children. For many other participants, there were noticeable feelings of loss and grief expressed. Participants were asked about whether any resources were available to help with education for decision-making or counseling on related psychosocial issues they have had related to decision-making. The most common response was that there were no notable resources available from an educational or counseling standpoint. Participants shared that the psychosocial challenges around decision-making brought up during the focus group were sensitive and personal issues, but nevertheless are topics worthy of discussion with healthcare professionals, whether it would be with providers with whom they have an established relationship or with genetic counselors.

**Perspectives on Genetic Counseling**

To our knowledge, this is the first study to explore perspectives on genetic counseling among individuals who have sickle cell disease. Three of the twenty-one
participants had received genetic counseling and spoke directly from their experiences. The other participants drew from a combination of what they knew about genetic counseling, the definition provided by the facilitator, and/or the experiences of fellow participants who shared their experiences directly. Overall, participants reported either having gained benefits or perceiving potential value from genetic counseling services. Individuals who received it said that they benefited from learning prenatal genetic risk information conveyed in a clear and understandable manner. Participants who had never heard about it or only knew about it broadly said that they saw it would be an appropriate source to learn more information about options available to them. Although individuals in this sample generally exhibited an understanding of the genetics behind sickle cell disease, several shared that they do not know all the reproductive options that are available to them and could benefit from learning about them in their future planning.

Most participants shared that they did not perceive any harm that could come directly from genetic counseling services. However, one of the three participants who had received genetic counseling shared a more critical perspective on her overall experience, mainly emphasizing that, despite reproductive risk being a sensitive and personal topic, the information provided by the genetic counselor felt simultaneously threatening and impersonal at the same time. This participant shared that she did not find the information useful and instead perceived that it was presented for the general masses. She had explained later that she felt as though a “wall” had already gone up as a result of feeling attacked, preventing her from being receptive to the information. Another of the three participants who received genetic counseling explained that she overall benefited from her genetic counseling. However, she recalled comments from the
clinician dissuading her from pregnancy; she conveyed that patients should be given autonomy over their reproductive decisions. These reports serve as reminders that risk information in the reproductive context is both sensitive and personal to patients. Effective, sensitive, and tailored communication of information is necessary to mitigate psychological and/or emotional harms that may occur from a genetic counseling discussion.

As discussed previously, participants in this current study emphasized the importance of their HCPs listening, effectively communicating, and building partnership with patients; these principles transfer to the genetic counseling context. Clinicians providing genetic counseling should ensure patients perceive that they have agency over the personal and family health information they provide, the risk information they receive, and any testing decisions they make. For this to happen, patients need to have a general context of what the genetic counseling discussion might entail as well as the opportunity to help set the agenda of the discussion to facilitate shared-decision making (Elwyn et al, 2000).

An overarching theme encompasses perceived barriers to genetic counseling. Concerns about exposure and access to genetic counseling were raised across the majority of the groups. Participants shared that young people likely would not know about the availability of genetic counseling or would have unanswered questions about access, like whether it would be covered by insurance or ways to locate genetic counseling services. Some insinuated how certain healthcare services like genetic counseling may not be as accessible for individuals from the Black/African-American population due to systematic or cultural influences. These findings highlight a potential
need for collaboration of genetics specialists, other healthcare professionals, and communities to determine and develop strategies that increase exposure to genetic counseling and lessen barriers of access.

Lastly, many of the participants in this study shared that they have strong working relationships with and high trust in their specialist providers. Some individuals shared that they would be more comfortable discussing the psychosocial issues related to reproductive decision-making brought up in focus groups with providers whom they know and trust. Because individuals with sickle cell disease meet with many different healthcare providers for comprehensive management, collaboration between genetics specialists and hematological specialist providers or primary care providers who they meet with more regularly may help patients feel the continuity of their care services.

**Strengths of Focus Groups**

Discussion with multiple participants simultaneously in a focus group setting can produce synergy, which occurs when interaction produces a combined greater effect than the sum of separate effects (i.e. individual interviews). The use of focus groups was ideal due to the nature of our study questions. Some of the discussion questions involved topics about which participants had little knowledge or experience (i.e. genetic counseling). The ability to reflect on each other’s ideas facilitated discussion. Some of the questions involved more sensitive topics, in which participants may have felt more comfortable sharing experiences among others who may have shared similar experiences.
Furthermore, focus groups are also often used in social science research that focuses on individuals of minority groups because they provide sensitivity to racial, ethnic, and cultural variables. In the United States, the African-American population is most affected by sickle cell disease. The goal was to provide a setting with a reduced researcher to participant ratio to provide more control over the discussion. This was particularly important to our study because of the difference between the racial identity of the facilitator and that of the participants. Furthermore, researchers have suggested that interviews in the group setting are more conducive to criticism of the health-care systems than individual interviews (Watts et al., 1987). This is likely facilitated by the synergy produced in a focus group setting. We noted that across all five focus groups of this current study, participants offered their candor regarding negative experiences as well as suggestions to address issues, such as lack of access to resources.

Study Limitations

The study has several limitations. Firstly, the participants were self-selected to participate in the focus group and were recruited primarily from the Baltimore-DC area. Some individuals were recruited from an advocacy organization and other individuals were recruited at the NIH, where they were already actively participating in other research studies. Furthermore, although this study did not ask participants about their highest attained education levels, we would characterize the sample to be well-educated relative to the general U.S. population. During the focus group discussions, many indicated that they have received bachelor’s degrees and/or post-graduate degrees and others indicated that they were in professions that require higher education. This sample,
therefore, is likely not representative of the broader population of individuals with SCD and results presented are not intended to be representative of the experiences and views of the entire population.

Additionally, perhaps the greatest risk of a focus group methodology is the potential for groupthink. This occurs when the responses of certain individuals lead others to subconsciously agree. Although the facilitator consistently asked participants for differing opinions during the discussion, there is little way to say definitively whether groupthink affected responses. Moreover, the racial discordance between the facilitator and participants could have influenced responses to questions on racial concordance and discordance in healthcare interactions. However, it is important to note a racially-concordant moderator would not necessarily have been guaranteed to elicit uninfluenced responses. Similarly, the moderator’s status as a genetic counseling graduate student at the time of the focus group discussions could have influenced responses regarding perceived benefits and harms from genetic counseling. In spite of these limitations, we were able to elicit criticisms of both healthcare relationships more generally and genetic counseling more specifically.

Furthermore, researchers have suggested that the ideal number of focus group participants ranges from six to ten participants to ensure a diversity of opinions and experiences. The number of participants in each group of this study averages to be just over four participants. However, we observed that our smaller groups yielded some benefits that mitigated the limitations of focus group research. With only one and a half hours to execute the focus group, the clearest benefit was that it allowed more time for each participant’s responses within the allocated time of the focus group. With smaller
groups, each participant’s responses are more fully represented in the data. The smaller group size also helped prevent any individuals from dominating the discussion. Participants also noted after the discussion that they considered the smaller size less overwhelming than previous larger forums. They mentioned that they felt more comfortable expressing disagreement in the smaller setting.

Furthermore, the participants in this study are all individuals with sickle cell disease and speak from their experiences with the condition. Although they spoke on behalf of African-American communities, their experiences, perspectives, and insights may not be representative of, for example, individuals with sickle cell trait. Although we do not claim the findings from this study alone are generalizable, findings may nonetheless inform foundations of future research and provide clinicians with some insight to better understand the experiences, needs, and barriers of patients living with sickle cell disease.

attitudes, and behaviors regarding sickle cell disease among college students.
Appendix A: Recruitment Scripts

Date: August 14, 2019
IRB Application: IRB00009643
PI: Debra Roter, PhD
Institution: Johns Hopkins School of Public Health

RECRUITMENT SCRIPTS

<table>
<thead>
<tr>
<th>Recruitment Script: In-person recruitment by DP (student researcher)</th>
</tr>
</thead>
</table>

For clinical settings of entities in which DP may be present for recruitment (i.e. NIH), a clinician who is associated with the clinical care or existing research study of the approached eligible individual asks for individual’s permission for DP to speak to them about a ~2-hour study that they are eligible for. If individual provides permission, DP will proceed to inform the individual of the study:

Hello, my name is Diana. I am a graduate student at Johns Hopkins. I am carrying out a research study on experiences with genetics services in healthcare among individuals from the sickle cell community. The purpose of the study is to help healthcare providers better understand experiences and opinions on genetics services from the SCD community to best deliver genetics services to patients like yourself. If you are an adult individual with sickle cell disease (*if recruitee is an adult with SCD*) OR a parent with sickle cell disease (*if recruitee is a parent of a child with SCD*), we invite you to participate. Are you interested in hearing more?

Are you interested in hearing more?

1) If “NO”: No problem at all. Thank you for your time!

2) If “YES”:

Great! Please see this recruitment flyer (Form: Recruitment Flyer) for the details in written form. Participating in this research is completely based on your choice (voluntary); you will not be faced with any penalty or denial of services if you do not join. Participation involves joining a focus group discussing with 5 to 7 other people
with sickle cell disease OR 5 to 7 other parents with sickle cell disease with a member of the research team leading the discussion. It will take around 1.5 hours. The discussion would be on your experiences of and opinions on genetics services. However, you do not have to have experience with genetics services to participate. Those who participate will get a $50-value gift card (choice of Amazon or Target). The focus group would occur at XXXX (time) on XXXX (date) at XXXXXXX (location) OR I will call you to plan a time and location for the focus group. Do you have questions about the study or about participation?

DP answers questions of the individual invited to the study.

Are you interested in participating?

1) If “NO”: No problem at all. Thank you for your time and have a good day!

2) If “YES”:

Wonderful! The flyer includes contact information the student researcher, me, can be reached at if you have questions or are interested later. If you know now you would like to participate, please fill this short form on your contact info and general availability (Form: Recruitee Contact and Availability). If you give permission, I will call you about scheduling and go over the consent information to make sure you are informed on the risks and benefits of the study. Please also take this demographic form (Form: Demographics and Survey Questionnaire) and bring it answered to the focus group.

OR (if time allows)

I’d like to go review this consent information with you to make sure you are informed on the risks and benefits of the study.

*DP reviews consent information, asks invited individual whether they have questions, and asks invited individual whether they would still like to participate.*

We ask that you complete short form on your availability and contact information. Please also take this demographic form (Form: Demographics and Survey Questionnaire) and bring it completed to the focus group if you participate. If you give permission, I will reach out to you about scheduling OR the focus group is located at XXXX location and at XXXX time on XXXX day.

Thank you!
Hi, we are working with a graduate student at Johns Hopkins carry out a research project. The project is to understand experiences with genetic services in healthcare among individuals from the sickle cell community. The purpose of the study is to help healthcare providers better understand experiences and opinions on genetics services from the SCD community to best deliver genetics services to patients like yourself. You are invited to participate in this study because you are an individual with sickle cell disease (if recruitee is an adult with SCD) OR a parent with sickle cell disease (if recruitee is a parent of a child with SCD). Are you interested in hearing more?

1) If “NO”: No problem at all. Thank you for your time!

2) If “YES”:

Great! Please see this recruitment flyer for the details in written form. Participating in this research is completely based on your choice (voluntary); you will not be faced with any penalty or denial of services if you do not join. Participation involves joining a focus group discussing with 5 to 7 other people with sickle cell disease OR 5 to 7 other parents with sickle cell disease with a member of the research team leading the discussion. It will take around 1.5 hours. The discussion would be on your experiences of and opinions on genetics services. Those who participate will get a $50-value gift card (choice of Amazon or Target). If recruiting for focus group that will be held at JHMI campus: You’ll be given a stamp to park in JHMI garages for free. The focus group would occur at XXXX (time) on XXXX (date) at XXXXXX (location) OR they will call you to plan a time and location for the focus group. Do you have any questions about the study or about participating?

Researcher answers any questions of individual invited to the study.

Are you interested in participating?

1) If “NO”: No problem at all. Thank you for your time and have a good day!

2) If “YES”:

The flyer includes contact information the student researcher can be reached at if you have questions or are interested later. If you know now you would like to participate and if we receive your permission to pass along your contact info, we will provide your contact info to the student researcher. Please fill out this short form on your contact info and general availability and the researcher will contact you (Form: Recruitee Contact and Availability). Please take this demographic form (Form: Demographics and Survey Questionnaire) and bring it answered to the focus group. Here is also a consent form for your reference. The student researcher will review this with you over-the-phone. She will also reach out to you about the focus group time and day.
I’d like to go review this consent information with you to make sure you are informed on the risks and benefits of the study. Individual assisting with recruitment reviews consent information, asks invited individual whether they have questions, and asks invited individual whether they would still like to participate.

Please take this consent information document for your reference. Please also take this demographic form (Form: Demographics and Survey Questionnaire) and bring it completed if you participate in the focus group. This flyer includes contact information the student researcher can be reached at if you have questions.

OR

Would you be OK with talking to a student researcher who can tell you more about the study?

If the individual gives permission to talk to the student in a nearby designated area outside the treating area, the student researcher will follow the script as follows.

“Thank you for your interest in participating in the study. The goal is to help understand opinions and experiences with genetics and genetics services from perspective of people with sickle cell disease and trait. You are not required to join as participation is on a volunteer basis. What it would look like to participate is joining a focus group discussion with 5-7 other individuals with sickle cell disease (or trait) for approximately an hour and a half. Participants will receive a 50$ gift card (choice of Amazon or Target) and free parking exit stamp (for groups at JHMI). (DP will either inform the individual of a scheduled focus group date or inform the individual that dates/times/locations will be chosen and offered to participants). The discussion groups will be in-person and there will be various locations, including at the Hopkins medical campus. Do you have any questions at this point?”

If the individual is still interested and asks questions, DP will answer questions. DP will then proceed to go over consent information on verbal consent document (and provide a physical copy) and allow the invited individual to ask questions. After answering questions (and if the individual agrees with the consent info), DP will ask the individual fill out information on the Recruitee Contact and General Availability form. DP will provide the Demographics and Survey Questionnaire form with instructions to bring it to the focus group.

Thank you!
Recruitment Script: over-the-phone recruitment by clinician or staff at recruitment site:

Hi __(name of recruitee being called)___, this is __(name and introduction of recruiting member)__. I am calling to tell you about a focus group research study you are eligible for. Is now an OK time to talk and are you in a private location? I expect it will take a little more than 5 minutes.

1) IF “NO”: No problem at all. Would you like me to call you back at another time?
2) IF “YES”:

Great! We are working with a graduate student at Johns Hopkins carry out a research project. The project is to understand experiences with genetic services in healthcare among individuals from the sickle cell community. The purpose of the study is to help healthcare providers better understand experiences and opinions on genetics services from the SCD community to best deliver genetics services to patients like yourself. You are invited to participate in this study because you are an individual with sickle cell disease (if recruitee is an adult with SCD) OR a parent with sickle cell disease (if recruitee is a parent of a child with SCD). Those who participate will get a $50-value gift card (choice of Amazon or Target). If recruiting for focus group that will be held at JHMI campus: You’ll be given a stamp to park in JHMI garages for free. Are you interested in hearing more?

1) IF “NO”: No problem at all. Thank you for your time!
2) If “YES”:

Participating in this research is completely based on your choice (voluntary); you will not be faced with any penalty or denial of services if you do not join. Participation involves joining an in-person focus group discussing with 5 to 7 other people with sickle cell disease OR 5 to 7 other parents with sickle cell disease with a member of the research team leading the discussion. It will take around 1.5 hours. The discussion would be on your experiences of and opinions on genetics services. However, you do not have to have experience with genetics services to participate.

Are you still interested?

1) If “NO”: No problem at all. Thank you for your time!
2) If YES: Great! Do you have any questions about the study or about participating?

Individual assisting with recruitment answers questions of individual invited to the study.
The focus group would occur at XXXX (time) on XXXX (date) at XXXXXX (location) OR the researcher will call you to plan a time and location for the focus group if you are okay with them being given your contact info.

If you are interested or have questions later, please reach out to the graduate student researcher at diana.phan@nih.gov or xxx-xxx-xxxx.
Hi (first name of participant)! Thank you for your interest in joining our study. My name is Diana, and I am the student researcher leading the study. Do you have questions you want to start off or would like general information about the research study.

1) IF “HAVE QUESTIONS”: I’d be happy to answer. Answer participant’s questions and provide remaining details about the study (see below).

2) IF “GENERAL INFORMATION”:

Absolutely. The project seeks to understand experiences with genetic services in healthcare among individuals from the sickle cell community. The purpose of the study is to help healthcare providers better understand experiences and opinions on genetics services from the SCD community to best deliver genetics services to patients like yourself. Any adult (18+ years old) who has sickle cell disease OR is a parent of a child with sickle cell disease and is fluent in English can choose to join.

Participation involves joining a 1.5-hour in-person focus group with 5-7 other people with sickle cell disease or other parents of children with sickle cell disease. The discussion would be on your experiences of and opinions on genetics services. Genetics services include genetic education, genetic testing, and genetic counseling. However, you do not need to have experience with genetics services to participate. Focus groups will occur in the Baltimore-DC area and will be assigned to the participant based on availability and location preferences. Participants will be given a $50-value gift card (choice of Amazon or Target). If recruiting for focus group that will be held at JHMI campus: You’ll be given a stamp to park in JHMI garages for free. The

Would you be interested in joining?

1) IF NO: No problem at all. Thanks for your time!

2) If “YES”:

Great! Could I please confirm with you the answers to the following questions?

- Are you 18 years of age or older?
- Do you speak English fluently?
- Do you have sickle cell disease? *
- Do you have a child with sickle cell disease? *

*Participants answering yes to either of these questions fit inclusion criteria if they answer yes to the two questions above.

Every research study comes with risks and benefits. I’d like to go over the consent information with you to make sure you understand the risks and benefits of this study. At the end I will ask you if you would still like to participate.
DP reviews consent information, asks invited individual whether they have questions, and asks invited individual whether they would still like to participate.

To assign focus group, we ask participants for availability and which location they prefer for focus groups. Ask questions on: Form: Recruitee Contact and Availability. OR the focus group will take place in XXXX location at XXXX time on XXXX day.

I would like to send it to you the consent document (Form: FocusGroup_Adult Oral Consent) we just reviewed along with a short survey we ask that you complete and bring to the focus group. Would you prefer the materials emailed to you or mailed to you physically? Please let me know an email address or mailing address.

Again, this research study is completely voluntary for you. If at any time you would not like to continue, please let me know as soon as you can. Thank you!
with SCD) OR a parent with a child who has sickle cell disease (if recruitee is a parent of a child with SCD), we invite you to participate. Are you interested in hearing more?

**Are you interested in hearing more?**

1) If “NO”: No problem at all. Thank you for your time!

2) If “YES”:

Great! Please see this recruitment flyer. Participation involves joining a focus group discussion with other people with sickle cell disease OR other parents with sickle cell disease. It will take around 1.5 hours. You do not have to have experience with genetics services (e.g. genetic testing, genetic counseling) to participate. Focus groups will occur in the Baltimore-DC area and will be assigned based on availability and location preferences. Those who participate will get a $50-value gift card (choice of Amazon or Target).

**Do you have any questions so far?**

*DP or AI answers questions accordingly.*

**Are you interested in participating?**

1) If “NO”: No problem at all. Thank you for your time and have a good day!

2) If “YES”:

**Screening: I first would like to ask a few questions to verify that you are eligible to participate in the study.**

Do you have sickle cell disease?

1) If “NO”: [Ask the next question]

2) If “YES”*

Are you a parent of a child with sickle cell disease?

1) If “NO”: Thank you for your time! We are only having the study with patients with sickle cell or their parents.

2) If “YES”*

Are you 18 years of age or older?
1) If “NO”: I’m sorry. We are only interviewing adults as part of her study.

2) If “YES”*: [Ask question about language.]

Is your primary language English or is it another language?

1) If “English”*: [Continue with script below]

2) If “another language”: Do you feel comfortable answering questions in English?
   a. If “YES”*: [Continue with script below]
   b. If “NO”: I’m sorry. We are only able to conduct the study in English at this time.

*Participants answering yes to these questions fit inclusion criteria if they answer in the affirmative to the other questions above.

**IF THE INDIVIDUAL IS ELIGIBLE:** Here is a Study Information Sheet that contains details about the study and participation. **DP or AI verbally covers the details on Study Information Sheet and asks potential participant if they have any questions. DP or AI covers multiple scenarios for scheduling and provides details: (1) focus group date(s) and time(s) already set and/or (2) future date and time will be scheduled based on availability.** **DP or AI will answer any questions accordingly.**

Are you still interested in participating?

If “YES”: [Continue with script below]

If “NO”: No problem. Thank you for your time and have a good day!

**IF FOCUS GROUP IS SCHEDULED:** Right now, we have a focus group scheduled at XXXX (time) on XXXX (date) at XXXXXX (location)]. Are you available to join us?

If "NO": [**DP or AI will provide alternate focus group dates if they are available**] OR If UNSURE OR NO FUTURE DATE IS SCHEDULED: Would you be willing to leave your contact information and general availability so that we can reach out to you in the future?

**IF "YES" TO A SPECIFIC FOCUS GROUP DATE:** **DP or the AI provides them any instructions/direction for focus group participation.** Here is a form asking about demographics that we ask you to complete and bring to the focus group. We would like to obtain your contact information so that we can reach out to you to remind you about the focus group. Please fill in this short form with your contact info and general availability (Recruitee Contact and Availability form).
IF "NO" TO DATE AND FUTURE CONTACT": [We are only offering this date and time.].
I thank you for your time and apologize for any inconvenience.

IF "YES" TO FUTURE CONTACT BUT NO DATE IS SELECTED: Here is a form asking about demographics that we ask you to complete and bring to the focus group. We would like to obtain your contact information so that we can reach out to you to schedule your participation in a focus group. Please fill in this short form with your contact info and general availability (Recruitee Contact and Availability form). We will contact you about scheduling a focus group.

Recruitment Script: over-the-phone recruitment by AI clinician or other AI at recruitment site:

Hi __ (name of recruitee being called) ___, this is __ (name and introduction of recruiting member) ___. I am calling to tell you about a focus group research study that you may be eligible for.

Is now an OK time to talk and are you in a private location? I expect it will take a little more than 5 minutes.

1) IF “NO”: No problem at all. Would you like me to call you back at another time?

2) IF “YES”:

Great! Researchers at NIH and Johns Hopkins are carrying out a research study on experiences with genetic services in healthcare among individuals from the sickle cell community. The purpose of the study is to help healthcare providers better understand experiences and opinions on genetics services from the SCD community to best deliver genetics services to patients. You are being invited to participate in this study because you are an individual with sickle cell disease (if recruitee is an adult with SCD) OR a parent with sickle cell disease (if recruitee is a parent of a child with SCD). Participation involves joining a focus group discussion with other people with sickle cell disease OR other parents with sickle cell disease. It will take around 1.5 hours. You do not have to have experience with genetics services (e.g. genetic testing, genetic counseling) to participate. Focus groups will occur in the Baltimore-DC area and will be assigned based on availability and location preferences. Those who participate will get a $50-value gift card (choice of Amazon or Target).

Are you interested in hearing more?

1) IF “NO”: No problem at all. Thank you for your time!
2) If “YES”: Do you have any questions so far?

Clinician answers questions accordingly.

Are you interested in participating?

1) If “NO”: No problem at all. Thank you for your time and have a good day!

2) If “YES”: (Only AI recruiting clinicians will offer to talk about next steps; non-AI clinicians will only gather contact info for researcher to contact potential participant.) Do you want to talk about next steps of the study (if clinician has time) or have another researcher contact you later about next steps? Talking about next steps now will take ten minutes or less.

IF NON-AI CLINICIAN OR “HAVE A RESEARCHER CONTACT ME LATER”: Clinician asks participant for permission to pass along contact information of individual to the researcher. Clinician asks for invited individual’s contact information and general availability using Recruitee Contact and Availability form.

IF “TALK ABOUT NEXT STEPS NOW”:

**Screening:** I first would like to ask a few questions to verify that you are eligible to participate in the study?

Do you have sickle cell disease?

1) If “NO”: [Ask the next question]

2) If “YES”*: [Ask question about age.]

Are you a parent of a child with sickle cell disease?

1) If “NO”: Thank you for your time! We are only interviewing patients with sickle cell or their parents.

2) If “YES”*: [Ask question about age.]

Are you 18 years of age or older?

1) If “NO”: I’m sorry. We are only interviewing adults as part of this study.

2) If “YES”*: [Ask question about language.]

Is your primary language English or is it another language?

3) If “ENGLISH”*: [Continue with script below]
4) If “another language”: Do you feel comfortable answering questions in English?

   a. If “YES”*: [Continue with script below]
   b. If “NO”: I’m sorry. We are only able to conduct the study in English at this time.

*Participants answering these questions fit inclusion criteria, if they answer in the affirmative to the other questions above.

IF THE INDIVIDUAL IS ELIGIBLE: The recruiting individual verbally covers the details on Study Information Sheet and asks potential participant if they have any questions. The recruiting individual covers multiple scenarios for scheduling and provides details: (1) focus group date(s) and time(s) already set and/or (2) future date and time will be scheduled based on availability. The recruiting individual will answer any questions accordingly.

Are you still interested in participating?

If “YES”: [Continue with script below]

If “NO”: No problem. Thank you for your time and have a good day!

IF FOCUS GROUP IS SCHEDULED: Right now, we have a focus group scheduled at XXXX (time) on XXXX (date) at XXXXXXX (location). Are you available to join us?

If "NO": [The recruiting individual will provide alternate focus group dates if they are available] OR If UNSURE OR NO FUTURE DATE IS SCHEDULED: Would you be willing to leave your contact information and general availability so that we can reach out to you in the future? Could I also email you the Study Information Sheet that contains details about the study and participation? (If individual responds with ‘yes,’ the recruiting individual completes the Recruitee Contact and Availability form and emails study information sheet.)

IF "YES" TO A SPECIFIC FOCUS GROUP DATE: The recruiting individual provides them any instructions/direction for focus group participation. Can I make sure that I have your correct contact info so that we can reach out to you to remind you about the focus group? (The recruiting individual completes the Recruitee Contact and Availability form.)

IF "NO" TO DATE AND FUTURE CONTACT": [We are only offering this date and time.]. I thank you for your time and apologize for any inconvenience.

IF "YES" TO FUTURE CONTACT BUT NO DATE IS SELECTED: We would like to confirm your contact information so that we can reach out to you to schedule your participation in a focus group. Can I make sure that I have your correct info and collect some additional information about your availability? (The recruiting individual completes the
Recruitee Contact and Availability form.) We will contact you about scheduling a focus group.

Could I email you the Study Information Sheet that contains details about the study and participation? I will also send you a form asking about demographics that we ask you to complete and bring to the focus group.

*If individual responds with ‘yes,’ the recruiting individual emails study information sheet and demographics form. If individual responds with ‘no,’ he or she will inform potential participant that they can be mailed the materials OR they will receive it at the focus group. In both scenarios, the recruiting individual will verbally state the details on Study Information Sheet and asks potential participant if they have any questions. The recruiting individual answers any questions accordingly.*

### Recruitment Script: Email from Clinician or Staff at recruitment site

Hello,

NIH and Johns Hopkins are carrying out a research study on experiences with genetic services in healthcare among individuals from the sickle cell community. The purpose of the study is to help healthcare providers better understand experiences and opinions on genetics services from the SCD community to best deliver genetics services to patients. You are invited to participate in this study if you are an individual with sickle cell disease OR a parent with sickle cell disease.

Participation is voluntary. It involves joining a 1.5-hour in-person focus group with 5-7 other people with sickle cell disease or other parents of children with sickle cell disease. The discussion would be on your experiences of and opinions on genetics services. Genetics services include genetic education, genetic testing, and genetic counseling. You do not need to have experience with genetics services to participate. Focus groups will occur in the Baltimore-DC area and will be assigned based on availability and location preferences. Participants will be given a $50-value gift card (choice of Amazon or Target).

Please see the attached flyer and Study Information Sheet for more information. If you are interested in participating or have questions, please reach out to diana.phan@nih.gov or XXX-XXX-XXXX.

Thank you!
<table>
<thead>
<tr>
<th>Recruitment and Screening Script: When Individuals Call DP with Interest in Participating or with Questions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hi (first name of participant)! Thank you for your interest in joining our study. My name is Diana, and I am the student researcher leading the study. Do you have questions you want to start off or would like general information about the research study?</td>
</tr>
</tbody>
</table>

1) **IF “HAVE QUESTIONS”:** I’d be happy to answer. Answer participant’s questions and provide remaining details about the study (see below).

2) **IF “GENERAL INFORMATION”:**

Absolutely. The project seeks to understand experiences with genetic services in healthcare among individuals from the sickle cell community. The purpose of the study is to help healthcare providers better understand experiences and opinions on genetics services from the SCD community to best deliver genetics services to patients. Any adult (18+ years old) who has sickle cell disease OR is a parent of a child with sickle cell disease and is fluent in English can choose to join.

Participation involves joining a 1.5-hour in-person focus group with 5-7 other people with sickle cell disease or other parents of children with sickle cell disease. The discussion would be on your experiences of and opinions on genetics services. Genetics services include genetic education, genetic testing, and genetic counseling. However, you do not need to have experience with genetics services to participate. Focus groups will occur in the Baltimore-DC area and will be assigned based on availability and location preferences. Participants will be given a $50-value gift card (choice of Amazon or Target).

**Do you have any questions so far?**

*DP answers questions accordingly.*

**Would you be interested in joining?**

1) **IF “NO”:** No problem at all. Thanks for your time! If you happen change your mind, please feel free to reach out to me again.

2) **If “YES”:**

**Screening:** I first would like to ask a few questions to verify that you are eligible to participate in the study?

Do you have sickle cell disease?
1) If “NO”: [Ask the next question]

2) If “YES”*: [Ask question about age.]

Are you a parent of a child with sickle cell disease?

1) If “NO”: Thank you for your time! We are only interviewing patients with sickle cell or their parents.

2) If “YES”*: [Ask question about age.]

Are you 18 years of age or older?

1) If “NO”: I’m sorry. We are only interviewing adults as part of this study.

2) If “YES”*: [Ask question about language.]

Is your primary language English or is it another language?

1) If “ENGLISH”*: [Continue with script below]

2) If “another language”: Do you feel comfortable answering questions in English?

   a. If “YES”*: [Continue with script below]

   b. If “NO”: I’m sorry. We are only able to conduct the study in English at this time.

*Participants answering these questions fit inclusion criteria, if they answer in the affirmative to the other questions above.

**IF THE INDIVIDUAL IS ELIGIBLE:** DP verbally covers the details on Study Information Sheet and asks potential participant if they have any questions. DP covers multiple scenarios for scheduling and provides details: (1) focus group date(s) and time(s) already set and/or (2) future date and time will be scheduled based on availability. DP will answer any questions accordingly.

Are you still interested in participating?

If “YES”: [Continue with script below]

If “NO”: No problem. Thank you for your time and have a good day!

**IF FOCUS GROUP IS SCHEDULED:** Right now, we have a focus group scheduled at XXXX (time) on XXXX (date) at XXXXXX (location). Are you available to join us?

If "NO": [DP will provide alternate focus group dates if they are available] OR IF UNSURE OR NO FUTURE DATE IS SCHEDULED: Would you be willing to leave your contact
information and general availability so that I can reach out to you in the future? Could I also email you the Study Information Sheet that contains details about the study and participation? *(If individual responds with 'yes,' DP completes the Recruitee Contact and Availability form and emails study information sheet.)*

**IF "YES" TO A SPECIFIC FOCUS GROUP DATE:** DP provides them any instructions/direction for focus group participation. Can I make sure that I have your correct contact info so that I can reach out to you to remind you about the focus group? *(DP completes the Recruitee Contact and Availability form.)*

**IF "NO" TO DATE AND FUTURE CONTACT**: [We are only offering this date and time.]. I thank you for your time and apologize for any inconvenience.

**IF "YES" TO FUTURE CONTACT BUT NO DATE IS SELECTED:** I would like to confirm your contact information so that we can reach out to you to schedule your participation in a focus group. Can I make sure that I have your correct info and collect some additional information about your availability? *(DP completes the Recruitee Contact and Availability form.)* I will contact you about scheduling a focus group.

Could I email you the Study Information Sheet that contains details about the study and participation? I will also send you a form asking about demographics that I ask you to complete and bring to the focus group.

*If individual responds with 'yes,' DP emails study information sheet and demographics form. If individual responds with 'no,' clinician will inform potential participant that they can be mailed the materials OR they will receive it at the focus group. In both scenarios, DP will verbally the details on Study Information Sheet and asks potential participant if they have any questions. DP answers any questions accordingly.*
Hello, I am Diana Phan from Johns Hopkins and would like to talk to you about genetics services for those with sickle cell disease.

- We are interested in improving genetics services (e.g. genetic counseling, genetic testing) for sickle cell disease. Understanding your experiences and opinions can help providers understand the needs and preferences of patients.
- We are asking adults with sickle cell disease and parents of children with sickle cell disease to participate in this research study. You do not have to join; it is your choice. There will be no penalty if you decide not to join.
- If you agree, you'll join a 90-minute discussion group with 5-7 other people with SCD or 5-7 other parents of children with SCD. The discussion will be audio-recorded and typed out for analysis by a company. At the end of the study we will also collect the questionnaire that we asked you to complete.
- You might feel uncomfortable answering some of the questions, but you do not have to answer every question and you do not have to share anything you do not wish to share.
- If you feel upset or distressed about the topics that come up in the discussion, we can give you information about organizations such as support groups and mental health service providers.
- There is a risk that someone outside the study will see your information. We will do our best to keep your information safe by not recording any identifiable information except for your voice on the audio recording.
- After the discussion, we will give you a $50-value gift card (your choice of Target or Amazon).
- We ask that you do not share information about your identity or information that could give hints on your identity. After the focus group, we also ask that you keep what is said in the conversation to yourself.

For questions and concerns:
- You may contact the principal investigator of the study, Debra Roter: o Phone: 410-955-9498
  o Email: droter1@jhu.edu
• Call or contact the **Johns Hopkins Bloomberg School of Public Health IRB Office** if you have questions about your rights as a study participant. Contact the IRB if you feel you have not been treated fairly or if you have other concerns. The IRB contact information is:

Telephone: 410-955-3193 Toll Free: 1-888-262-3242
E-mail: jhsph.irboffice@jhu.edu

**STUDY INFORMATION SHEET: FOCUS GROUP DISCUSSION ON GENETICS SERVICES**

**What is this study about?**
Thank you for your interest in our research study! The goal of this study is to explore experiences and opinions of patients and the parents of patients to help improve genetics services (e.g. genetic counseling, genetic testing) for sickle cell disease. Understanding your experiences and opinions can help genetics providers understand the needs and preferences of patients like yourself.

**What are the requirements to participate?**
To participate, you must be 18 years or older and be able to speak English fluently. You must also either have a diagnosis of sickle cell disease OR have at least one child with a diagnosis of sickle cell disease.

**What will I be asked to do if I agree to participate?**
You will be asked to participate in a group discussion with one trained interviewer and 5-7 other people with SCD or other parents of children with SCD. The focus group will take approximately 90-minutes to complete.

We will ask about your general experiences as individuals with SCD or parents of children with SCD. We will also ask about your experiences with and opinions on genetics services. When it comes to answering questions, there are no right or wrong answers. We just want to know what you think.
What is the payment for participation?
You will receive a $50-value gift card (choice of Amazon or Target) at the end of your participation.

Would my participation in this study be kept confidential?
The researchers will do everything we can to protect your identity and what you say during the focus group. Research data will be stored in a password-protected computer files. When we store your data, we will take precautions to protect your information from others that should not have access to it. For example, when appropriate, we will remove information that can identify you and use a code instead.

What are the risks of this research?
We do not think that being part of this discussion will cause harm to you. However, all research comes with risks. We cannot guarantee that other focus group participants will keep discussions confidential, but we will remind and encourage all participants to maintain high respect for privacy. You do not have to answer any questions that you do not want to. If a topic during the discussion causes you to feel upset or uncomfortable, you do not have to continue with the study. If this happens, we have contact information of organizations that we can provide to you.

Even with the protections we put in place, we cannot absolutely guarantee that your identity will never become known and in a rare case, someone could still gain access to your identifiable information.

What are the benefits of this research?
There are no direct benefits to participating in this research study. However, we hope that your experiences and opinions could teach healthcare professionals how to improve genetics services for patients like yourself or your child.

Do I have to be in this research and may I stop participating at any time?
Your participation in this research is completely voluntary, and you may change your mind at any time if you do decide to participate. If you decide not to participate in this activity or if you stop participating at any time, you will not be penalized.

How do I proceed with the study and ask questions?
Please contact the lead researcher (contact information below), who will answer questions and explain next steps. If you are interested the team will do their best to work with your schedule to include you in the study.

Diana Phan  
Email: diana.phan@nih.gov  
Phone: 301-402-5184
Appendix C: Contact and Availability Form

RECRUITEE CONTACT AND GENERAL AVAILABILITY FORM

Protocol Name: Voices from the Sickle Cell Community on Genetics Services: A Qualitative Exploration

Recruitee Name (Full first and initial of last name): ____________________________________

Phone Number: ________________________________________________________________

Email: ________________________________________________________________________

Prefer to be reached by: phone or email

Recruitee generally available for focus group participation (check all that apply):

Days of Week:

<table>
<thead>
<tr>
<th>Mon</th>
<th>Tues</th>
<th>Wed</th>
<th>Thurs</th>
<th>Fri</th>
<th>Sat</th>
<th>Sun</th>
</tr>
</thead>
</table>

Times:

<table>
<thead>
<tr>
<th>Mornings (8AM-12PM)</th>
<th>Early Afternoons (12PM-3PM)</th>
<th>Late Afternoons (3PM-6PM)</th>
<th>Evenings (6PM-9PM)</th>
</tr>
</thead>
</table>

I Prefer to attend a focus group discussion in:

BALTIMORE

WASHINGTON DC

Notes from Recruitee (if applicable):
Appendix D: Demographics and Survey Questionnaire

1. Which age range do you fall under?

<table>
<thead>
<tr>
<th>Age Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>18-25 years old</td>
</tr>
<tr>
<td>26-35 years old</td>
</tr>
<tr>
<td>36-45 years old</td>
</tr>
<tr>
<td>46-55 years old</td>
</tr>
<tr>
<td>56-65 years old</td>
</tr>
<tr>
<td>66-75 years old</td>
</tr>
<tr>
<td>76+</td>
</tr>
</tbody>
</table>

2. Which race do you identify as (please select all that apply; continued on back)?

<table>
<thead>
<tr>
<th>Race</th>
</tr>
</thead>
<tbody>
<tr>
<td>White</td>
</tr>
<tr>
<td>Black or African American</td>
</tr>
<tr>
<td>American Indian or Alaska Native</td>
</tr>
<tr>
<td>Asian</td>
</tr>
<tr>
<td>Native Hawaiian or Other Pacific Islander</td>
</tr>
<tr>
<td>Other (please specify):</td>
</tr>
</tbody>
</table>

3. Which ethnicity do you identify as?

<table>
<thead>
<tr>
<th>Ethnicity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hispanic or Latino(a)</td>
</tr>
<tr>
<td>Not Hispanic or Latino</td>
</tr>
</tbody>
</table>

4. Which gender do you identify as?

<table>
<thead>
<tr>
<th>Gender</th>
</tr>
</thead>
<tbody>
<tr>
<td>female</td>
</tr>
</tbody>
</table>

_______________________________________________________________________

5. Do you have sickle cell disease? (please circle one):

<table>
<thead>
<tr>
<th>YES</th>
<th>NO</th>
</tr>
</thead>
</table>

6. Do you have children?
7. Do you have a child or children with sickle cell disease?
   YES  NO

8. If YES to the above question, please write the number of children you have who
   have sickle cell disease and list their ages: ________________________________

9. For each of your children with sickle cell disease, list how old they were when
   you learned that they had sickle cell disease.
   ________________________________________________________________
   (examples: during pregnancy, shortly after birth, age 3)

10. Have you met with any type of genetic counselor or other genetic specialist
    before? (please circle one):
        YES  NO

Appendix E: Focus Group Facilitator Guide

1) Go over Study Information Sheet/Verbal Consent Form with participants

2) Introduction to Focus Group:
a. Thank participants for participating in focus group discussion. DP explains the significance of the focus group and how it could contribute to improving genetic healthcare services. DP introduces herself as the moderator and researcher.

b. Explain that the discussion will be driven by the participants and will be between the participants, and that the moderator will only pose questions and help the conversation flow. Explain that participant does not have to answer every question and does not have to answer any question.

c. Ask participant to state their assigned ID code prior to speaking (e.g. “B speaking” or “AA speaking”)

3) Icebreaker: Have participants introduce themselves and one fun fact

**Adults with SCD:**

Q: If you were to briefly explain what it means to have sickle cell disease to someone who doesn’t know the condition, what would you share?

Q: A lot of clinicians aren’t familiar with sickle cell disease. What advice would you give clinicians to better understand the experiences of a parent with sickle cell disease?

  - Probe for responses on how sickle cell disease is different for each person who has it: What advice would you give to clinicians to understand how different sickle cell disease is for each person who has it?

Q: What are some concerns that you had when planning to have children?

  - Probe for personal health-related concerns (e.g. potentially shorter life span, pregnancy complications): Could you share more about the concerns you had with your health?
  - Probe for concerns about passing off sickle cell disease to children: Could you share more about your concerns about passing on sickle cell disease to your children?

Follow-up Q: What resources were helpful during this time? What resources could have been helpful when you were planning a family?

  - Probe for informational and emotional support resources
  - Probe for helpful resources from community

Q: Has a clinician, family member, or someone from your community ever explained to you how a person gets sickle cell disease? What was your experience?
- Probe for perceptions on the information delivered: Was information correct and clear? Was the information helpful? Was there enough information given? Did the clinician use confusing medical terms (jargon)?
- Probe for genetic counseling experiences → explore that experience further: You mentioned you met with a genetic counselor. How would you describe that experience?
- Does anyone have a different experience?

Introduce role of genetic counselor: A genetic counselor is a healthcare professional who helps with some of the topics we discussed. They provide information and resources so patients can understand a genetic condition and the genetics behind the conditions. The information is to help patients make decisions about their health, about genetic testing and about family planning based on the patient’s preferences and values. Genetic counselors can also help families consider how to best talk about sickle cell disease with other family members or help people learn ways of coping with the condition. Genetic counselors often see adults with SCD and SCT before or during pregnancy. They also sometimes see adults outside of pregnancy or parents of children found to have SCD and SCT.

Q: Has anyone had experience with genetic counseling before?

Q: What are your thoughts on genetic counseling?
   - Probe for both perceived benefits and harms: What was helpful about genetic counseling? What was not helpful? What did you not like about the experience?

Q: Are there ways in which a genetic counselor can be helpful to you now?

Q: If your children were to see a genetic counselor, what would you want the genetic counselor to keep in mind to be most helpful for your kids?
   - Probe for communication skills, sensitivity to stigma, or racial and genetic discrimination → “could you share more about this?”

Follow-up (if applicable per earlier discussion): Would the race/culture/gender/age of the genetic counselor matter?

Given the earlier discussion related to stigma, how would you want genetic counselors to change what they do? (then you can probe for: any changes to how they communicate with people? Any changes to the ways in which they talk about sickle cell disease?

Q: How do you think society sees sickle cell disease?
- Probe for lack of awareness: Could you share more of your thoughts on how not many people know about sickle cell disease?
- Probe for racialized beliefs: Could you explain more about how you think there are stereotypes about race and sickle cell disease?

Q: What causes sickle cell disease? Who gets sickle cell disease? What groups are at risk for getting sickle cell disease?

Q: You have been talking about who YOU think is at risk for sickle cell disease. In your experience, what do other people in your community think about who is at risk for developing sickle cell? What do you think healthcare professionals think about this?

Q: does it matter than this difference in understanding exists in your community? In the country? In the medical community? If so, how? If not, why doesn’t it matter?

Follow-up Q: There is a common incorrect belief that sickle disease only occurs in African American or Black individuals. What are your thoughts on this?

Follow-up Q: How much do healthcare professionals (who aren’t familiar with sickle cell disease) think that sickle cell disease is a “Black disease”?
   - Probe for perceived outcomes of this misbelief in the medical community: Do you think that this stereotype or misconception has positive or negative consequences?

Q: Does anyone know scientifically why the “sickle cell gene” exists?

Scientists have discovered that sickle cell trait happened in humans over 7,000 years ago. Sickle cell trait and sickle cell disease still exist today because people with trait have stronger protection from the deadly infection malaria. While the trait gene started in Africa, both the trait and disease can be found many parts of the world among many ethnicities. This is because of people migrating throughout history. Scientists say the sickle cell trait gene protected the world from malaria.

Follow-up Q: What are your thoughts on this information? Does it change anything for you? If so, what? If not, why not? Would it have changed anything if you had heard this earlier in your life? If so, what would have changed? If not, why not?

Follow-up Q: How helpful would it be if clinicians were to tell people this information?

Follow-up Q: Other thoughts?
REFERENCES


Adejumo, A. O., & Olaoye, O. M. (2018). Moderating role of gender on genetics knowledge, perceived need for genetic testing and attitude to genetic counseling among people living with SCD. Gender and Behaviour, 16(1), 11152-11167.


Braun, L. (2002). Race, ethnicity, and health: can genetics explain disparities?. Perspectives in Biology and Medicine, 45(2), 159-174.


Rotimi, C. N. (2004). Are medical and nonmedical uses of large-scale genomic markers conflating genetics and 'race'?. Nature genetics, 36(11s), S43.


Williams-Smith, M. (2015). Factors that contribute to the knowledge, health beliefs,
CURRICULUM VITAE

Diana Phan
12903 Standish Dr., Poway, CA 92064
(858) 231- 3151 • dianaphan94@gmail.com

EDUCATION

Johns Hopkins University & National Institutes of Health, MD
Department of Health, Behavior, and Society, Johns Hopkins Bloomberg School of Public Health
Medical Genetics and Metabolic Genomics Branch, National Human Genome Research Institute & National Cancer Institute, NIH
- Master of Science in Genetic Counseling • GPA: 3.94 • March 2020
- Relevant coursework: Cancer Genetics, Human Genetics, Medical Genetics and Genomic Medicine, Health Decision-Making, Biostatistics, Epidemiology

University of California, Santa Barbara, CA
Department of Molecular, Cellular, and Developmental Biology
- Bachelor of Science in Biological Sciences • GPA: 3.7 (Honors) • June 2016
- Relevant coursework: Cell Growth and Oncogenesis, Biochemistry, Human Genetics, Human Physiology

Poway High School, CA
- June 2012

AWARDS & HONORS

National Human Genome Research Institute, NIH
Pre-doctoral Intramural Research Training Award Fellowship • 2017-2020
- Awarded as part of acceptance to the Johns Hopkins University/National Institutes of Health Genetic Counseling Training Program to support research training and endeavors

University of California, Santa Barbara
Dean’s Honors • 2012-2016
- Awarded for academic achievement in coursework

University of California, Santa Barbara
Regents Scholarship • 2012-2016
- $24,000 merit-based academic scholarship awarded by the University of California to entering undergraduates

College Board
Advanced Placement Scholar with Distinction • 2012
- Awarded for achievements on college-level coursework and exams during high school
GENETIC COUNSELING TRAINING ROTATIONS

**Johns Hopkins Center for Inherited Heart Disease, MD**
Genetic Counseling Trainee • September 2019 - present
- Provide supervised genetic risk assessment, counseling, and testing in addition to medical screening and management guidance to patients and their families on inherited heart conditions (i.e. arrhythmia, cardiomyopathy, and hypercholesteremia conditions with known inherited bases)
- Responsible for managing cases comprehensively at both the clinical and administrative level (i.e. documentation of notes/patient letters, ordering genetic tests, returning over-the-phone or in-person test results, and addressing patient insurance issues)

**National Cancer Institute, Neuro-Oncology Branch, MD**
Genetic Counseling Trainee • September 2019 – October 2019
- Provided supervised genetic risk assessment, counseling, and testing to research participants in studies for rare neuro-oncological tumors (i.e. astrocytoma, meningioma, and ependymoma)
- Obtained and documented participants’ phenotypic data for research and assessments of risk for inherited conditions/cancer syndromes
- Observed management and physical evaluation visits with neuro-oncologists
- Observed genetic counseling for research participants with history of gastrointestinal stromal tumors

**National Human Genome Research Institute, Medical Genomics and Metabolic Genetics Branch, MD**
Genetic Counseling Trainee • June 2019 – July 2019
- Provided supervised genetic risk assessment, counseling, and testing to research participants and their families in a natural history study for methylmalonic acidemia and cobalamin disorders and propionic acidemia, inborn errors, or organic acid metabolism
- Developed patient-friendly educational materials on metabolic diseases for research participants and their families

**Johns Hopkins Comprehensive Cancer Care Center, MD**
Genetic Counseling Trainee • April 2019 – June 2019
- Provided supervised genetic risk assessment, counseling, and testing to patients at-risk for hereditary cancer predisposition syndromes
- Interpreted patients’ somatic genetic testing results to recommend germline counseling and testing appropriately
- Observed screening and management counseling by genetic oncologists (Drs. Frank Giardiello and Kala Visvanathan)

**Johns Hopkins DNA Diagnostic Laboratory, MD**
Genetic Counseling Trainee • October 2018 – December 2018
- Carried out variant classification and exome-level analysis as well as clinical report writing for those classifications
• Analyzed CFTR variants for the Cystic Fibrosis Mutation Analysis Program, which provides free genetic testing for individuals symptomatic for CF who have not had genetic testing
• Curated a list of genes associated with inherited metabolic conditions that is being used for the laboratory’s “Zoom” exome panels

**Johns Hopkins Prenatal Diagnosis and Treatment, MD**
Genetic Counseling Trainee • September 2018 – October 2018
• Provided supervised genetic risk assessment, counseling, and testing for high-risk pregnancies (e.g. advanced maternal age, abnormal ultrasound and bloodwork, family history or known carrier status of genetic condition, multiple miscarriages)
• Assisted with ordering of genetic tests and documented visit notes
• Delivered results counseling both over-the-phone and in-person

**Kennedy Krieger Institute, Department of Neurology, MD**
Genetic Counseling Trainee • July 2018 – September 2018
• Provided supervised genetic counseling and testing to adult and pediatric patients and their families in the neurogenetics, leukodystrophy, and muscle dystrophy clinics
• Developed patient-friendly resources for patients and families to better understand complicated and/or uncertain genetic findings
• Documented visit notes
• Observed evaluation and management carried out by neurologists

**Walter Reed Medical Institute, General Genetics, MD**
Genetic Counseling Trainee • March 2018 – May 2018
• Provided supervised genetic counseling and testing to active military patients and their families
• Assisted with determining/confirming diagnoses through genetic testing and testing for carrier status
• Assisted with selection of genetic testing panels and documented visit notes
• Counseled patients on potential implications of genetic testing on military employment
• Observed evaluation and management carried out by a medical geneticist

**Mercy Medical Center, Center for Advanced Fetal Care, MD**
Genetic Counseling Trainee • January 2018 – March 2018
• Provided supervised reproductive genetic counseling and testing to patients with pregnancies at-risk for genetic or chromosomal conditions
• Assisted with ordering of tests and documentation of physician letters

**National Center for Advancing Translational Sciences, Genetic and Rare Diseases Information Center, MD**
Genetic Counseling Trainee • September 2017– October 2017
• Responded to online inquiries on genetic and rare diseases from individuals and their families from across the United States and internationally
Provided patrons with detailed summaries of the medical literature translated to laymen’s terms, online resources, contact information of advocacy groups, and providers based on the patron’s needs and geographic locations

Created webpage content for rarediseases.info.nih.gov, specifically on Rahman Syndrome

LEADERSHIP & VOLUNTEERING

**Johns Hopkins Bloomberg School of Public Health**
**Vice President of Student Groups, Student Assembly • Sept. 2018- June 2019**
- Served on the leadership board of Student Assembly and participating in formulation of policies and regulations
- Acted as a liaison between student groups and the Bloomberg School administration
- Responsible for administrative management of over 40 student groups/organizations to ensure compliance with school’s requirements for active status
- Planned and organized annual volunteer and recreational events for students of Johns Hopkins University and Medicine

**Johns Hopkins University & National Institutes of Health, MD**
**Volunteer, DNA Day • April 2019**
- Visited biology classrooms of Baltimore High Schools and lead modules for students on pharmacogenetics and DNA extraction
- Introduced the career of genetic counseling for students and shared ways for students who are interest to enter or learn more about the field

**University of California, Santa Barbara**
**President, Regents and Chancellor’s Scholars Association • Sept. 2015- June 2016**
- Coordinate events and opportunities for students to network with the university chancellor and faculty members
- Provided oversight of the association’s treasurer, secretary, and communication chair
- Planned and organized community service events for members of the association and university students

**University of California, Santa Barbara**
**Volunteer Group Leader, SciTrek • 2014**
- Lead modules for 5th grade students in Santa Barbara that include hands-on chemistry and physics experiments
- Taught students age-appropriate basics of the scientific method

**University of California, Santa Barbara**
**Speaker Liaison and Event Coordinator, TEDxUCSB • 2013**
- Event Theme: “Energy to Power: Harnessing the Potential of Today for Tomorrow”
- Helped with selection of speakers and acted as a speaker liaison before and during the event
Co-planned, organized, and publicized TEDx event which was attended by over 150 university students, staff, and faculty

**RESEARCH**

**Johns Hopkins University & National Institutes of Health, MD**

Master’s Thesis: Shaping a Dialogue: Insights from the Sickle Cell Community on Genetics Healthcare

Principal Investigators: Debra Roter, DrPH and Lori Erby, PhD, ScM, CGC

- **Objective:** Several studies and professional organizations promote genetic counseling for individuals with sickle cell disease and trait. Because genetic testing within the sickle cell context has historically been tensioned by racial discrimination and stigma in the U.S., we argue for the importance of exploring current opinions, experiences, and concerns of the community to inform ways to enhance the benefits and mitigate the harms of genetic counseling for individuals affected with SCT and SCD. The present study queries perspectives of affected adults and unaffected parents on genetic counseling and related issues as well as perspectives on racialized vs ancestral views of SCD in the medical and broader communities.

- **Methods:** Carrying out qualitative focus group discussions with a convenience sample of 30-40 adults with sickle cell disease parents of children with sickle cell disease who are recruited from 5 sites in the Baltimore-DC area. Qualitatively analyzing the data for themes and subthemes related to the research questions stated above.

**University of California, Santa Barbara, CA**

Undergraduate researcher, Weimbs Laboratory • 2013-2015

Advisor: Thomas Weimbs, PhD

- Lab focus: Autosomal Dominant Polycystic Kidney Disease (ADPKD)
- Worked closely with post-doctoral fellow to investigate the role of kinase PKCzeta in ADPKD. PKCzeta has been found by the Weimbs Lab to interact directly with polycystin-1, suggesting that it could play a role in

**University of California, Santa Barbara, CA**

Undergraduate research assistance, Girvetz School of Education • 2015-2016

Advisor: Meghan Corella, PhD

- Assisted a graduate student on a qualitative study that examines the role of use of academic language in the elementary classroom setting
- Roles included transcribing and coding audio of observational videos of the classroom setting

**PRESENTATIONS**

**National Institutes of Health, MD**

National Human Genome Research Institute Annual Symposium • November 2019

- Poster Presentation Title: “Shaping a Dialogue: Insights from the Sickle Cell Community on Genetics Healthcare”
- Audience: training fellows, staff, and employees of the National Human Genome Research Institute

National Institutes of Health, MD
Post-Clinic Conference, National Human Genome Research Institute
- Title: “Implications of Genetic Test Results on Life Insurers’ Decisions: The Current Landscape”
- Audience: medical genetics fellows, post-doctoral researchers, and faculty clinicians and scientists of the National Human Genome Research Institute

Johns Hopkins Medicine, MD
Sickle cell Improvement across the NorthEast ReGion through Education
Research Meeting • October 2019
- Title: “Insights from the Sickle Cell Community on Genetics Healthcare” (master’s thesis preliminary findings)
- Audience: clinicians, researchers, and medical students interested in sickle cell disease care

Johns Hopkins Comprehensive Cancer Center, MD
Genetics Case Conference • May 2019
- Title: “Implications of Genetic Test Results on Life Insurers’ Decisions: The Current Landscape”
- Audience: cancer genetic counselors, medical oncologists, clinic staff

Johns Hopkins DNA Diagnostic Lab
- Title: “A Curation of Genes Associated with Inherited Metabolic Disease and Inborn Errors of Metabolism”
- Audience: laboratory genetic counselors, scientific laboratory director, research staff

National Institutes of Health, MD
Post-Clinic Conference, National Human Genome Research Institute • February 2018
- Title: “Psychosocial Implications of Receiving Variants of Uncertain Cancer Genetic Test Results”
- Audience: medical genetics fellows, post-doctoral researchers, and faculty clinicians and scientists of the National Human Genome Research Institute

CONFERENCES ATTENDED
- National Society of Genetic Counselor Annual Conference (2018 & 2019)
- Sickle Cell Disease Association of America Annual National Convention (2019)
- Maryland Sickle Cell Disease Association (2019)

WORK EXPERIENCE
Self-employed, San Diego County
Private Tutor • July 2016 – May 2017
- Tutored 20+ students on a weekly basis in math, chemistry, biology, and SAT/ACT standardized tests
- Student grade levels ranged from middle school to high school
University of California, Santa Barbara, CA
Group and Individual Chemistry Tutor, Campus Learning Assistance Services (CLAS) • Sept. 2015- June 2016
- Created and implemented lesson plans to tutor undergraduates in general chemistry
- Taught study skills and tactics to help students better understand concepts, problem-solving, and improve at test-taking
- Maintained communication with professors of coursework tutored to optimize tutoring lessons for students
- Individually tutored student athletes and students

University of California, Santa Barbara, CA
Administrative Assistant, Weimbs Laboratory • 2010-2011
- Manage laboratory website and online safety systems for lab members
- Order equipment and materials for laboratory
- Manage travel scholarship information of graduate student/postdoctoral fellows of the university’s molecular, cellular, and developmental biology department